Gareth Evans

List of Publications by Year in Descending Order

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	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
954	Rare germline copy number variants (CNVs) and breast cancer risk.. *Communications Biology*, **2022**, 5, 65	6.7	0
953	Naevoid basal cell carcinoma syndrome **2022**, 449-452		
952	Women's health behaviour change after receiving breast cancer risk estimates with tailored screening and prevention recommendations.. *BMC Cancer*, **2022**, 22, 69	4.8	2
951	Common variants in breast cancer risk loci predispose to distinct tumor subtypes.. *Breast Cancer Research*, **2022**, 24, 2	8.3	3
950	Pathology of Tumors Associated With Pathogenic Germline Variants in 9 Breast Cancer Susceptibility Genes.. *JAMA Oncology*, **2022**,	13.4	4
949	Beyond Antoni: A Surgeon's Guide to the Vestibular Schwannoma Microenvironment.. *Journal of Neurological Surgery, Part B: Skull Base*, **2022**, 83, 1-10	1.5	1
948	The importance of ethnicity: Are breast cancer polygenic risk scores ready for women who are not of White European origin?. *International Journal of Cancer*, **2022**, 150, 73-79	7.5	1
947	Dominant-negative pathogenic variant BRIP1 c.1045G>C is a high-risk allele for non-mucinous epithelial ovarian cancer: A case-control study. *Clinical Genetics*, **2022**, 101, 48-54	4	0
946	Abstract P1-10-01: Results from the breast cancer - anti progestin prevention study 1 (BC-APPS1) trial - a novel approach in breast cancer prevention. *Cancer Research*, **2022**, 82, P1-10-01-P1-10-01	10.1	
945	Earlier decisions on breast and ovarian surgery reduce cancer in women at high risk.. *BMJ, The*, **2022** , 376, o258	5.9	0
944	Risk perception and disease knowledge in attendees of a community-based lung cancer screening programme.. *Lung Cancer*, **2022**, 168, 1-9	5.9	0
943	A Genome-Wide Gene-Based Gene–Environment Interaction Study of Breast Cancer in More than 90,000 Women. *Cancer Research Communications*, **2022**, 2, 211-219		0
942	Solving the genetic aetiology of hereditary gastrointestinal tumour syndromes- a collaborative multicentre endeavour within the project Solve-RD.. *European Journal of Medical Genetics*, **2022**, 104475	2.6	0
941	Genome-wide and transcriptome-wide association studies of mammographic density phenotypes reveal novel loci.. *Breast Cancer Research*, **2022**, 24, 27	8.3	1
940	The feasibility of implementing risk stratification into a national breast cancer screening programme: a focus group study investigating the perspectives of healthcare personnel responsible for delivery.. *BMC Women's Health*, **2022**, 22, 142	2.9	1
939	Does receiving high or low breast cancer risk estimates produce a reduction in subsequent breast cancer screening attendance? Cohort study.. *Breast*, **2022**, 64, 47-49	3.6	0

938	Breast cancer risks associated with missense variants in breast cancer susceptibility genes.. *Genome Medicine*, **2022**, 14, 51	14.4	0
937	Personalised Risk Prediction in Hereditary Breast and Ovarian Cancer: A Protocol for a Multi-Centre Randomised Controlled Trial. *Cancers*, **2022**, 14, 2716	6.6	1
936	The importance of genetic counseling and screening for people with pathogenic SMARCE1 variants: A family study. *American Journal of Medical Genetics, Part A*, **2021**, 185, 561-565	2.5	1
935	GORLIN SYNDROME **2021**, 459-474		0
934	A prospective prostate cancer screening programme for men with pathogenic variants in mismatch repair genes (IMPACT): initial results from an international prospective study. *Lancet Oncology, The*, **2021**, 22, 1618-1631	21.7	10
933	Analysis of the Li-Fraumeni Spectrum Based on an International Germline TP53 Variant Data Set: An International Agency for Research on Cancer TP53 Database Analysis. *JAMA Oncology*, **2021**,	13.4	4
932	Age at diagnosis of cancer in 185delAG BRCA1 mutation carriers of diverse ethnicities: tentative evidence for modifier factors. *Familial Cancer*, **2021**, 20, 189-194	3	
931	Assessment of mismatch repair deficiency in ovarian cancer. *Journal of Medical Genetics*, **2021**, 58, 687-693	5.8	2
930	Constitutional de novo deletion CNV encompassing predisposes to diffuse hyperplastic perilobar nephroblastomatosis (HPLN). *Journal of Medical Genetics*, **2021**, 58, 581-585	5.8	0
929	Disease course of neurofibromatosis type 2: a 30-year follow-up study of 353 patients seen at a single institution. *Neuro-Oncology*, **2021**, 23, 1113-1124	1	3
928	Germline FFPE inherited cancer panel testing in deceased family members: implications for clinical management of unaffected relatives. *European Journal of Human Genetics*, **2021**, 29, 861-871	5.3	0
927	Extended gene panel testing in lobular breast cancer. *Familial Cancer*, **2021**, 1	3	
926	Prospective evaluation of a breast-cancer risk model integrating classical risk factors and polygenic risk in 15 cohorts from six countries. *International Journal of Epidemiology*, **2021**,	7.8	6
925	Identifying challenges in neurofibromatosis: a modified Delphi procedure. *European Journal of Human Genetics*, **2021**, 29, 1625-1633	5.3	1
924	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. *Genetics in Medicine*, **2021**, 23, 1779-1782	8.1	2
923	Current recommendations for cancer surveillance in Gorlin syndrome: a report from the SIOPE host genome working group (SIOPE HGWG). *Familial Cancer*, **2021**, 20, 317-325	3	6
922	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	7.5	2
921	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. *Genetics in Medicine*, **2021**, 23, 1506-1513	8.1	43

920	Extending screening intervals for women at low risk of breast cancer: do they find it acceptable?. *BMC Cancer*, **2021**, 21, 637	4.8	0
919	Cognitive and Electrophysiological Correlates of Working Memory Impairments in Neurofibromatosis Type 1. *Journal of Autism and Developmental Disorders*, **2021**, 1	4.6	2
918	PTCH2 is not a strong candidate gene for gorlin syndrome predisposition. *Familial Cancer*, **2021**, 1	3	2
917	Microscopy and chemical analyses reveal flavone-based woolly fibres extrude from micron-sized holes in glandular trichomes of Dionysia tapetodes. *BMC Plant Biology*, **2021**, 21, 258	5.3	0
916	Clinical utility of testing for PALB2 and CHEK2 c.1100delC in breast and ovarian cancer. *Genetics in Medicine*, **2021**, 23, 1969-1976	8.1	1
915	The Relationship between Body Mass Index and Mammographic Density during a Premenopausal Weight Loss Intervention Study. *Cancers*, **2021**, 13,	6.6	2
914	A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. *European Journal of Human Genetics*, **2021**, 29, 1354-1358	5.3	5
913	The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. *Genetics in Medicine*, **2021**, 23, 1726-1737	8.1	2
912	No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in and : A Prospective Lynch Syndrome Database Study. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
911	Patient reported outcome measures in a cohort of patients at high risk of breast cancer treated by bilateral risk reducing mastectomy and breast reconstruction. *Journal of Plastic, Reconstructive and Aesthetic Surgery*, **2021**,	1.7	2
910	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. *European Journal of Human Genetics*, **2021**, 29, 1337-1347	5.3	4
909	Genotype-Phenotype Correlations in Neurofibromatosis and Their Potential Clinical Use. *Neurology*, **2021**, 97, S91-S98	6.5	5
908	Breast cancer incidence and early diagnosis in a family history risk and prevention clinic: 33-year experience in 14,311 women. *Breast Cancer Research and Treatment*, **2021**, 189, 677-687	4.4	1
907	Combined Associations of a Polygenic Risk Score and Classical Risk Factors With Breast Cancer Risk. *Journal of the National Cancer Institute*, **2021**, 113, 329-337	9.7	14
906	NF1 optic pathway glioma: analyzing risk factors for visual outcome and indications to treat. *Neuro-Oncology*, **2021**, 23, 100-111	1	9
905	Sporadic vestibular schwannoma: a molecular testing summary. *Journal of Medical Genetics*, **2021**, 58, 227-233	5.8	2
904	Specialist oncological surgery for removal of the ovaries and fallopian tubes in BRCA1 and BRCA2 pathogenic variant carriers may reduce primary peritoneal cancer risk to very low levels. *International Journal of Cancer*, **2021**, 148, 1155-1163	7.5	5
903	A mismatch in care: results of a United Kingdom-wide patient and clinician survey of gynaecological services for women with Lynch syndrome. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2021**, 128, 728-736	3.7	8

902	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	7.5	2
901	, a gene for colorectal cancer predisposition in the absence of Li-Fraumeni-associated phenotypes. *Gut*, **2021**, 70, 1139-1146	19.2	3
900	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	8.1	9
899	Early Adaptation of Colorectal Cancer Cells to the Peritoneal Cavity Is Associated with Activation of "Stemness" Programs and Local Inflammation. *Clinical Cancer Research*, **2021**, 27, 1119-1130	12.9	1
898	Lynch syndrome for the gynaecologist. *The Obstetrician and Gynaecologist*, **2021**, 23, 9-20	0.9	10
897	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. *Human Mutation*, **2021**, 42, 223-236	4.7	29
896	The spatial phenotype of genotypically distinct meningiomas demonstrate potential implications of the embryology of the meninges. *Oncogene*, **2021**, 40, 875-884	9.2	3
895	Neuroimaging manifestations in children with SARS-CoV-2 infection: a multinational, multicentre collaborative study. *The Lancet Child and Adolescent Health*, **2021**, 5, 167-177	14.5	81
894	Preventing Ovarian Cancer through early Excision of Tubes and late Ovarian Removal (PROTECTOR): protocol for a prospective non-randomised multi-center trial. *International Journal of Gynecological Cancer*, **2021**, 31, 286-291	3.5	6
893	European guidelines from the EHTG and ESCP for Lynch syndrome: an updated third edition of the Mallorca guidelines based on gene and gender. *British Journal of Surgery*, **2021**, 108, 484-498	5.3	30
892	Attitudes towards risk-reducing early salpingectomy with delayed oophorectomy for ovarian cancer prevention: a cohort study. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2021**, 128, 714-726	3.7	6
891	Advances in genetic technologies result in improved diagnosis of mismatch repair deficiency in colorectal and endometrial cancers. *Journal of Medical Genetics*, **2021**,	5.8	1
890	Targeting lung cancer screening to individuals at greatest risk: the role of genetic factors. *Journal of Medical Genetics*, **2021**, 58, 217-226	5.8	3
889	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. *British Journal of Cancer*, **2021**, 124, 842-854	8.7	2
888	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
887	Current recommendations for clinical surveillance and genetic testing in rhabdoid tumor predisposition: a report from the SIOPE Host Genome Working Group. *Familial Cancer*, **2021**, 20, 305-316	3	5
886	Choose and stay on one out of two paths: distinction between clinical versus research genetic testing to identify cancer predisposition syndromes among patients with cancer. *Familial Cancer*, **2021**, 20, 289-291	3	0
885	Comment on: SMARCB1 Gene Mutation Predisposes to Earlier Development of Glioblastoma: A Case Report of Familial GBM. *Journal of Neuropathology and Experimental Neurology*, **2021**, 80, 289-290	3.1	1

884	The Angelina Jolie effect: Contralateral risk-reducing mastectomy trends in patients at increased risk of breast cancer. *Scientific Reports*, **2021**, 11, 2847	4.9	5
883	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
882	Surgical decision making in premenopausal carriers considering risk-reducing early salpingectomy or salpingo-oophorectomy: a qualitative study. *Journal of Medical Genetics*, **2021**,	5.8	2
881	Survival from breast cancer in women with a BRCA2 mutation by treatment. *British Journal of Cancer*, **2021**, 124, 1524-1532	8.7	2
880	High likelihood of actionable pathogenic variant detection in breast cancer genes in women with very early onset breast cancer. *Journal of Medical Genetics*, **2021**,	5.8	3
879	Pathogenic noncoding variants in the neurofibromatosis and schwannomatosis predisposition genes. *Human Mutation*, **2021**, 42, 1187-1207	4.7	1
878	Oral contraceptive use and ovarian cancer risk for BRCA1/2 mutation carriers: an international cohort study. *American Journal of Obstetrics and Gynecology*, **2021**, 225, 51.e1-51.e17	6.4	9
877	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. *Journal of the National Cancer Institute*, **2021**,	9.7	3
876	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. *Lancet Oncology, The*, **2021**, 22, 1014-1022	21.7	5
875	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. *Breast Cancer Research*, **2021**, 23, 86	8.3	1
874	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. *British Journal of Cancer*, **2021**, 125, 1135-1145	8.7	0
873	Implementation of Multigene Germline and Parallel Somatic Genetic Testing in Epithelial Ovarian Cancer: SIGNPOST Study. *Cancers*, **2021**, 13,	6.6	8
872	Gene Panel Testing for Breast Cancer Reveals Differential Effect of Prior Probability. *Cancers*, **2021**, 13,	6.6	1
871	Lessons learned from drug trials in neurofibromatosis: A systematic review. *European Journal of Medical Genetics*, **2021**, 64, 104281	2.6	0
870	Translabyrinthine resection of NF2 associated vestibular schwannoma with cochlear implant insertion. *Neurosurgical Focus Video*, **2021**, 5, V14	0.1	
869	Uptake of bilateral-risk-reducing-mastectomy: Prospective analysis of 7195 women at high-risk of breast cancer. *Breast*, **2021**, 60, 45-52	3.6	4
868	Breast Cancer Risk Factors and Survival by Tumor Subtype: Pooled Analyses from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2021**, 30, 623-642	4	4
867	Optical coherence tomography significance in managing complex neurofibromatosis 2-related papilledema: Report of a case. *JRSM Open*, **2021**, 12, 2054270420981454	0.5	

866	From to Polygenic Risk Scores: Mutation-Associated Risks in Breast Cancer-Related Genes. *Breast Care*, **2021**, 16, 202-213	2.4	1
865	Introducing a low-risk breast screening pathway into the NHS Breast Screening Programme: Views from healthcare professionals who are delivering risk-stratified screening. *Womenś Health*, **2021**, 17, 17455065211009746	3	2
864	Testing a breast cancer prevention and a multiple disease prevention weight loss programme amongst women within the UK NHS breast screening programme-a randomised feasibility study.. *Pilot and Feasibility Studies*, **2021**, 7, 220	1.9	0
863	Germline Testing in Breast Cancers: Why, When and How?. *Cancers*, **2020**, 12,	6.6	7
862	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
861	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. *Nature*, **2020**, 583, 90-95	50.4	69
860	Predictors of long-term cancer-related distress among female BRCA1 and BRCA2 mutation carriers without a cancer diagnosis: an international analysis. *British Journal of Cancer*, **2020**, 123, 268-274	8.7	11
859	Psychosocial effects of whole-body MRI screening in adult high-risk pathogenic mutation carriers: a case-controlled study (SIGNIFY). *Journal of Medical Genetics*, **2020**, 57, 226-236	5.8	8
858	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. *Human Mutation*, **2020**, 41, 1555-1562	4.7	9
857	Disease expression in juvenile polyposis syndrome: a retrospective survey on a cohort of 221 European patients and comparison with a literature-derived cohort of 473 SMAD4/BMPR1A pathogenic variant carriers. *Genetics in Medicine*, **2020**, 22, 1524-1532	8.1	11
856	Response to Benusiglio et al. *Genetics in Medicine*, **2020**, 22, 1424-1425	8.1	1
855	Guidelines for the Li-Fraumeni and heritable TP53-related cancer syndromes. *European Journal of Human Genetics*, **2020**, 28, 1379-1386	5.3	61
854	Characterising the loss-of-function impact of 5' untranslated region variants in 15,708 individuals. *Nature Communications*, **2020**, 11, 2523	17.4	35
853	Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. *European Journal of Human Genetics*, **2020**, 28, 1387-1393	5.3	27
852	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
851	Li-Fraumeni Exploration Consortium Data Coordinating Center: Building an Interactive Web-Based Resource for Collaborative International Cancer Epidemiology Research for a Rare Condition. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 927-935	4	3
850	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. *Journal of Medical Genetics*, **2020**, 57, 829-834	5.8	11
849	Personalized early detection and prevention of breast cancer: ENVISION consensus statement. *Nature Reviews Clinical Oncology*, **2020**, 17, 687-705	19.4	64

848	Feasibility of Gynaecologist Led Lynch Syndrome Testing in Women with Endometrial Cancer. *Journal of Clinical Medicine*, **2020**, 9,	5.1	5
847	Prostate Cancer Risk by BRCA2 Genomic Regions. *European Urology*, **2020**, 78, 494-497	10.2	2
846	Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled trial. *Lancet, The*, **2020**, 395, 1855-1863	40	88
845	What are the benefits and harms of risk stratified screening as part of the NHS breast screening Programme? Study protocol for a multi-site non-randomised comparison of BC-predict versus usual screening (NCT04359420). *BMC Cancer*, **2020**, 20, 570	4.8	16
844	Long-Term Evaluation of a UK Community Pharmacy-Based Weight Management Service. *Pharmacy (Basel, Switzerland)*, **2020**, 8,	2	3
843	Young adulthood body mass index, adult weight gain and breast cancer risk: the PROCAS Study (United Kingdom). *British Journal of Cancer*, **2020**, 122, 1552-1561	8.7	7
842	European women's perceptions of the implementation and organisation of risk-based breast cancer screening and prevention: a qualitative study. *BMC Cancer*, **2020**, 20, 247	4.8	9
841	The inflammatory microenvironment in vestibular schwannoma. *Neuro-Oncology Advances*, **2020**, 2, vdaa023	0.3	13
840	Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *JAMA Oncology*, **2020**, 6, 1218-1230	13.4	25
839	Risk of Contralateral Breast Cancer in Women with and without Pathogenic Variants in , and Genes in Women with Very Early-Onset (. *Cancers*, **2020**, 12,	6.6	11
838	Neurofibromatosis type 2 discordance in monozygous twins. *Familial Cancer*, **2020**, 19, 37-40	3	2
837	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for and Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2020**, 29, 368-378	4	9
836	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
835	Risk-reducing salpingo-oophorectomy, natural menopause, and breast cancer risk: an international prospective cohort of BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2020**, 22, 8	8.3	22
834	Perceived fatigue in children and young adults with neurofibromatosis type 1. *Journal of Paediatrics and Child Health*, **2020**, 56, 878-883	1.3	8
833	Association between genetic polymorphisms and endometrial cancer risk: a systematic review. *Journal of Medical Genetics*, **2020**, 57, 591-600	5.8	15
832	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
831	The introduction of risk stratified screening into the NHS breast screening Programme: views from British-Pakistani women. *BMC Cancer*, **2020**, 20, 452	4.8	6

830	Screening Strategy Modification Based on Personalized Breast Cancer Risk Stratification and its Implementation in the National Guidelines - Pilot Study. *Zdravstveno Varstvo*, **2020**, 59, 211-218	1.3	
829	Clinical and neuroradiological characterisation of spinal lesions in adults with Neurofibromatosis type 1. *Journal of Clinical Neuroscience*, **2020**, 77, 98-105	2.2	2
828	Prostate Cancer Risks for Male BRCA1 and BRCA2 Mutation Carriers: A Prospective Cohort Study. *European Urology*, **2020**, 77, 24-35	10.2	53
827	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
826	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
825	Risk-based breast cancer screening strategies in women. *Best Practice and Research in Clinical Obstetrics and Gynaecology*, **2020**, 65, 3-17	4.6	20
824	Engagement barriers and service inequities in the NHS Breast Screening Programme: Views from British-Pakistani women. *Journal of Medical Screening*, **2020**, 27, 130-137	1.4	10
823	Use of anastrozole for breast cancer prevention (IBIS-II): long-term results of a randomised controlled trial. *Lancet, The*, **2020**, 395, 117-122	40	54
822	Hereditary Leiomyomatosis and Renal Cell Cancer: Clinical, Molecular, and Screening Features in a Cohort of 185 Affected Individuals. *European Urology Oncology*, **2020**, 3, 764-772	6.7	13
821	Sporadic implementation of UK familial mammographic surveillance guidelines 15 years after original publication. *British Journal of Cancer*, **2020**, 122, 329-332	8.7	1
820	Challenging the believed proportion of ovarian cancer attributable to BRCA2 versus BRCA1 pathogenic variants. *European Journal of Cancer*, **2020**, 124, 88-90	7.5	1
819	Genetic predisposition to cancer. *Medicine*, **2020**, 48, 138-143	0.6	
818	Mammographic density change in a cohort of premenopausal women receiving tamoxifen for breast cancer prevention over 5 years. *Breast Cancer Research*, **2020**, 22, 101	8.3	5
817	Regarding "Neuro-Oncology Practice Clinical Debate: targeted therapy vs conventional chemotherapy in pediatric low-grade glioma". *Neuro-Oncology Practice*, **2020**, 7, 572-573	2.2	0
816	Preferences for breast cancer prevention among women with a or mutation. *Hereditary Cancer in Clinical Practice*, **2020**, 18, 20	2.3	1
815	Mainstreaming germline BRCA1/2 testing in non-mucinous epithelial ovarian cancer in the North West of England. *European Journal of Human Genetics*, **2020**, 28, 1541-1547	5.3	11
814	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1653-1666	8.1	34
813	Infantile fibrosarcoma with TPM3-NTRK1 fusion in a boy with Bloom syndrome. *Familial Cancer*, **2020**, 1	3	0

812	BRCA1 and BRCA2 pathogenic variant carriers and endometrial cancer risk: A cohort study. *European Journal of Cancer*, **2020**, 136, 169-175	7.5	12
811	Risk stratified breast cancer screening: UK healthcare policy decision-making stakeholders' views on a low-risk breast screening pathway. *BMC Cancer*, **2020**, 20, 680	4.8	7
810	Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database. *Journal of Clinical Medicine*, **2020**, 9,	5.1	7
809	Future Research Suggestions for Multigene Testing in Unselected Populations-Reply. *JAMA Oncology*, **2020**, 6, 785-786	13.4	
808	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 837-848	11	12
807	The proportion of endometrial tumours associated with Lynch syndrome (PETALS): A prospective cross-sectional study. *PLoS Medicine*, **2020**, 17, e1003263	11.6	19
806	Reply to Kratz et al. *European Journal of Human Genetics*, **2020**, 28, 1483-1485	5.3	3
805	Long-Term Evaluation of Women Referred to a Breast Cancer Family History Clinic (Manchester UK 1987-2020). *Cancers*, **2020**, 12,	6.6	2
804	The microenvironment in sporadic and neurofibromatosis type II-related vestibular schwannoma: the same tumor or different? A comparative imaging and neuropathology study. *Journal of Neurosurgery*, **2020**, 134, 1419-1429	3.2	8
803	Epithelial ovarian cancer risk: A review of the current genetic landscape. *Clinical Genetics*, **2020**, 97, 54-63	4	18
802	A deep intronic SMARCB1 variant associated with schwannomatosis. *Clinical Genetics*, **2020**, 97, 376-377	4	5
801	Incidence of mosaicism in 1055 de novo NF2 cases: much higher than previous estimates with high utility of next-generation sequencing. *Genetics in Medicine*, **2020**, 22, 53-59	8.1	27
800	A case-control evaluation of 143 single nucleotide polymorphisms for breast cancer risk stratification with classical factors and mammographic density. *International Journal of Cancer*, **2020**, 146, 2122-2129	7.5	19
799	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
798	Breast cancer in neurofibromatosis 1: survival and risk of contralateral breast cancer in a five country cohort study. *Genetics in Medicine*, **2020**, 22, 398-406	8.1	15
797	EANO guideline on the diagnosis and treatment of vestibular schwannoma. *Neuro-Oncology*, **2020**, 22, 31-45	1	66
796	Germline and sporadic cancers driven by the RAS pathway: parallels´and´contrasts. *Annals of Oncology*, **2020**, 31, 873-883	10.3	14
795	Uptake of pre-symptomatic testing for and is age, gender, offspring and time-dependent. *Journal of Medical Genetics*, **2020**,	5.8	2

794	UKCGG Consensus Group guidelines for the management of patients with constitutional pathogenic variants. *Journal of Medical Genetics*, **2020**,	5.8	7
793	Autism Spectrum Disorder Symptom Profile Across the RASopathies. *Frontiers in Psychiatry*, **2020**, 11, 585700	5	5
792	Re-evaluating genetic variants identified in candidate gene studies of breast cancer risk using data from nearly 280,000 women of Asian and European ancestry. *EBioMedicine*, **2019**, 48, 203-211	8.8	9
791	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	2.3	16
790	Cost-effectiveness analysis of reflex testing for Lynch syndrome in women with endometrial cancer in the UK setting. *PLoS ONE*, **2019**, 14, e0221419	3.7	12
789	Two truncating variants in FANCC and breast cancer risk. *Scientific Reports*, **2019**, 9, 12524	4.9	2
788	Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. *European Urology*, **2019**, 76, 831-842	10.2	78
787	A Cost-effectiveness Analysis of Multigene Testing for All Patients With Breast Cancer. *JAMA Oncology*, **2019**,	13.4	46
786	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019** , 10, 431	17.4	45
785	Prevalence of germline pathogenic variants in sequential epithelial ovarian cancer cases. *Journal of Medical Genetics*, **2019**, 56, 301-307	5.8	12
784	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. *Human Mutation*, **2019**, 40, 1557-1578	4.7	52
783	An alternative approach to establishing unbiased colorectal cancer risk estimation in Lynch syndrome. *Genetics in Medicine*, **2019**, 21, 2706-2712	8.1	6
782	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2019**, 121, 180-192	8.7	13
781	Multiple primary malignancies associated with a germline SMARCB1 pathogenic variant. *Familial Cancer*, **2019**, 18, 445-449	3	1
780	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
779	Confirmation that somatic mutations of beta-2 microglobulin correlate with a lack of recurrence in a subset of stage II mismatch repair deficient colorectal cancers from the QUASAR trial. *Histopathology*, **2019**, 75, 236-246	7.3	8
778	The proportion of endometrial cancers associated with Lynch syndrome: a systematic review of the literature and meta-analysis. *Genetics in Medicine*, **2019**, 21, 2167-2180	8.1	69
777	Trends in phenotype in the English paediatric neurofibromatosis type 2 cohort stratified by genetic severity. *Clinical Genetics*, **2019**, 96, 151-162	4	9

776	Lifestyle behaviours and health measures of women at increased risk of breast cancer taking chemoprevention. *European Journal of Cancer Prevention*, **2019**, 28, 500-506	2	2
775	Cancer surveillance, obesity, and potential bias. *Lancet Public Health, The*, **2019**, 4, e218	22.4	
774	Comparison of a Standard Resolution PET-CT Scanner With an HRRT Brain Scanner for Imaging Small Tumors Within the Head. *IEEE Transactions on Radiation and Plasma Medical Sciences*, **2019**, 3, 434-443	4.2	4
773	Breast cancer in patients with germline TP53 pathogenic variants have typical tumour characteristics: the Cohort study of TP53 carrier early onset breast cancer (COPE study). *Journal of Pathology: Clinical Research*, **2019**, 5, 189-198	5.3	15
772	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
771	Concern regarding classification of germline TP53 variants as likely pathogenic. *Human Mutation*, **2019**, 40, 828-831	4.7	5
770	A Micro-Costing Study of Screening for Lynch Syndrome-Associated Pathogenic Variants in an Unselected Endometrial Cancer Population: Cheap as NGS Chips?. *Frontiers in Oncology*, **2019**, 9, 61	5.3	8
769	Women's perceptions of personalized risk-based breast cancer screening and prevention: An international focus group study. *Psycho-Oncology*, **2019**, 28, 1056-1062	3.9	16
768	The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. *Genetics in Medicine*, **2019**, 21, 2390-2400	8.1	79
767	Neurosurgical contribution within a complex NF1 supraregional service. *Clinical Neurology and Neurosurgery*, **2019**, 180, 18-24	2	2
766	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	2.3	24
765	Breast cancer pathology and stage are better predicted by risk stratification models that include mammographic density and common genetic variants. *Breast Cancer Research and Treatment*, **2019**, 176, 141-148	4.4	30
764	Genome-wide association study of germline variants and breast cancer-specific mortality. *British Journal of Cancer*, **2019**, 120, 647-657	8.7	28
763	Phase 0 trial investigating the intratumoural concentration and activity of sorafenib in neurofibromatosis type 2. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 1184-1187	5.5	3
762	The association between weight at birth and breast cancer risk revisited using Mendelian randomisation. *European Journal of Epidemiology*, **2019**, 34, 591-600	12.1	11
761	Final Results of the Prospective FH02 Mammographic Surveillance Study of Women Aged 35-39 at Increased Familial Risk of Breast Cancer. *EClinicalMedicine*, **2019**, 7, 39-46	11.3	5
760	Sarcoma in neurofibromatosis 2: case report and review of the literature. *Familial Cancer*, **2019**, 18, 97-100	1.9	1
759	Psychosocial impact of undergoing prostate cancer screening for men with BRCA1 or BRCA2 mutations. *BJU International*, **2019**, 123, 284-292	5.6	7

758	Development of Breast Cancer Choices: a decision support tool for young women with breast cancer deciding whether to have genetic testing for BRCA1/2 mutations. *Supportive Care in Cancer*, **2019**, 27, 297-309	3.9	6
757	Lynch syndrome screening in gynaecological cancers: results of an international survey with recommendations for uniform reporting terminology for mismatch repair immunohistochemistry results. *Histopathology*, **2019**, 75, 813-824	7.3	9
756	Comparative performances of machine learning methods for classifying Crohn Disease patients using genome-wide genotyping data. *Scientific Reports*, **2019**, 9, 10351	4.9	44
755	The genetic interplay between body mass index, breast size and breast cancer risk: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2019**, 48, 781-794	7.8	16
754	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
753	Prediction of reader estimates of mammographic density using convolutional neural networks. *Journal of Medical Imaging*, **2019**, 6, 031405	2.6	16
752	Gliomas in the context of Li-Fraumeni syndrome: An international cohort.. *Journal of Clinical Oncology*, **2019**, 37, 1517-1517	2.2	4
751	Distinct Immunological Landscapes Characterize Inherited and Sporadic Mismatch Repair Deficient Endometrial Cancer. *Frontiers in Immunology*, **2019**, 10, 3023	8.4	18
750	Predictors of weight gain in a cohort of premenopausal early breast cancer patients receiving chemotherapy. *Breast*, **2019**, 45, 1-6	3.6	9
749	Identifying modifiable and non-modifiable risk factors of epithelial ovarian cancer□an we get it better?. *Gynecology and Pelvic Medicine*, **2019**, 2, 21-21	0.3	
748	[18F]fluorothymidine and [18F]fluorodeoxyglucose PET Imaging Demonstrates Uptake and Differentiates Growth in Neurofibromatosis 2 Related Vestibular Schwannoma. *Otology and Neurotology*, **2019**, 40, 826-835	2.6	4
747	Neurofibromatosis type 2 and related disorders. *Current Opinion in Oncology*, **2019**, 31, 562-567	4.2	12
746	Breast cancer risk status influences uptake, retention and efficacy of a weight loss programme amongst breast cancer screening attendees: two randomised controlled feasibility trials. *BMC Cancer*, **2019**, 19, 1089	4.8	10
745	Results of multigene panel testing in familial cancer cases without genetic cause demonstrated by single gene testing. *Scientific Reports*, **2019**, 9, 18555	4.9	9
744	Rapid reversal of clinical down-classification of a BRCA1 splicing variant avoiding psychological harm. *Clinical Genetics*, **2019**, 95, 532-533	4	1
743	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
742	Beliefs About Medication and Uptake of Preventive Therapy in Women at Increased Risk of Breast Cancer: Results From a Multicenter Prospective Study. *Clinical Breast Cancer*, **2019**, 19, e116-e126	3	13
741	European Breast Cancer Council manifesto 2018: Genetic´risk prediction testing in breast cancer. *European Journal of Cancer*, **2019**, 106, 45-53	7.5	14

740	Genetic counselling and testing of susceptibility genes for therapeutic decision-making in breast cancer-an European consensus statement and expert recommendations. *European Journal of Cancer*, **2019**, 106, 54-60	7.5	13
739	Global Disparities in Breast Cancer Genetics Testing, Counselling and Management. *World Journal of Surgery*, **2019**, 43, 1264-1270	3.3	7
738	Are women with pathogenic variants in PMS2 and MSH6 really at high lifetime risk of breast cancer?. *Genetics in Medicine*, **2019**, 21, 1878-1879	8.1	3
737	Association Between Invasive Lobular Breast Cancer and Mutations in the Mismatch Repair Gene MSH6. *JAMA Oncology*, **2019**, 5, 119-120	13.4	
736	Inflammation and vascular permeability correlate with growth in sporadic vestibular schwannoma. *Neuro-Oncology*, **2019**, 21, 314-325	1	29
735	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. *Journal of the National Cancer Institute*, **2019**, 111, 350-364	9.7	22
734	Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. *Familial Cancer*, **2019**, 18, 281-284	3	6
733	Familial unilateral vestibular schwannoma is rarely caused by inherited variants in the NF2 gene. *Laryngoscope*, **2019**, 129, 967-973	3.6	9
732	Breast cancer risk in neurofibromatosis type 1 is a function of the type of gene mutation: a new genotype-phenotype correlation. *Journal of Medical Genetics*, **2019**, 56, 209-219	5.8	19
731	Identifying the deficiencies of current diagnostic criteria for neurofibromatosis 2 using databases of 2777 individuals with molecular testing. *Genetics in Medicine*, **2019**, 21, 1525-1533	8.1	28
730	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2019**, 48, 795-806	7.8	52
729	Neurofibromatosis type 2 service delivery in England. *Neurochirurgie*, **2018**, 64, 375-380	1.4	10
728	Population-based testing of non-mucinous epithelial ovarian cancer in Scotland. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2018**, 125, 1459	3.7	1
727	Penetrance estimates for , (also applied to Lynch syndrome) based on presymptomatic testing: a new unbiased method to assess risk?. *Journal of Medical Genetics*, **2018**, 55, 442-448	5.8	1
726	PARP inhibitors in platinum-sensitive high-grade serous ovarian cancer. *Cancer Chemotherapy and Pharmacology*, **2018**, 81, 647-658	3.5	47
725	Findings Linking Mismatch Repair Mutation With Age at Endometrial and Ovarian Cancer Onset in Lynch Syndrome-Reply. *JAMA Oncology*, **2018**, 4, 890-891	13.4	0
724	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
723	Use of Single-Nucleotide Polymorphisms and Mammographic Density Plus Classic Risk Factors for Breast Cancer Risk Prediction. *JAMA Oncology*, **2018**, 4, 476-482	13.4	72

722	Evaluation of the relative effectiveness of the 2017 updated Manchester scoring system for predicting mutations in a Southeast Asian country. *Journal of Medical Genetics*, **2018**, 55, 344-350	5.8	3
721	Psychosocial issues of a population approach to high genetic risk identification: Behavioural, emotional and informed choice issues. *Breast*, **2018**, 37, 148-153	3.6	11
720	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
719	Systematic review of the empirical investigation of resources to support decision-making regarding BRCA1 and BRCA2 genetic testing in women with breast cancer. *Patient Education and Counseling*, **2018**, 101, 779-788	3.1	13
718	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
717	Cost effectiveness of population based BRCA1 founder mutation testing in Sephardi Jewish women. *American Journal of Obstetrics and Gynecology*, **2018**, 218, 431.e1-431.e12	6.4	23
716	Prostate-specific antigen velocity in a prospective prostate cancer screening study of men with genetic predisposition. *British Journal of Cancer*, **2018**, 118, 266-276	8.7	9
715	Randomised controlled trial of simvastatin treatment for autism in young children with neurofibromatosis type 1 (SANTA). *Molecular Autism*, **2018**, 9, 12	6.5	33
714	Germline pathogenic variants in PALB2 and other cancer-predisposing genes in families with hereditary diffuse gastric cancer without CDH1 mutation: a whole-exome sequencing study. *The Lancet Gastroenterology and Hepatology*, **2018**, 3, 489-498	18.8	58
713	Personalized prevention in high risk individuals: Managing hormones and beyond. *Breast*, **2018**, 39, 139-147	3.6	15
712	Diagnosis of sporadic neurofibromatosis type 2 in the paediatric population. *Archives of Disease in Childhood*, **2018**, 103, 463-469	2.2	16
711	Risks of breast or ovarian cancer in BRCA1 or BRCA2 predictive test negatives: findings from the EMBRACE study. *Genetics in Medicine*, **2018**, 20, 1575-1582	8.1	12
710	A Novel PTCH1 Frameshift Mutation Leading to Nevoid Basal Cell Carcinoma Syndrome. *Cytogenetic and Genome Research*, **2018**, 154, 57-61	1.9	5
709	Are we ready for the challenge of implementing risk-based breast cancer screening and primary prevention?. *Breast*, **2018**, 39, 24-32	3.6	19
708	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
707	High-Grade Glioma is not a Feature of Neurofibromatosis Type 2 in the Unirradiated Patient. *Neurosurgery*, **2018**, 83, 193-196	3.2	7
706	An update on the diagnosis and treatment of vestibular schwannoma. *Expert Review of Neurotherapeutics*, **2018**, 18, 29-39	4.3	44
705	CNVs affecting cancer predisposing genes (CPGs) detected as incidental findings in routine germline diagnostic chromosomal microarray (CMA) testing. *Journal of Medical Genetics*, **2018**, 55, 89-96	5.8	5

704	Malignant Peripheral Nerve Sheath Tumors are not a Feature of Neurofibromatosis Type 2 in the Unirradiated Patient. *Neurosurgery*, **2018**, 83, 38-42	3.2	12
703	A Dominantly Inherited 5' UTR Variant Causing Methylation-Associated Silencing of BRCA1 as a Cause of Breast and Ovarian Cancer. *American Journal of Human Genetics*, **2018**, 103, 213-220	11	43
702	NFM-04. INITIAL MANAGEMENT STRATEGY AS A DISCRIMINATOR OF VISUAL OUTCOME IN CHILDREN PRESENTING WITH NEUROFIBROMATOSIS TYPE 1 AND OPTIC PATHWAY GLIOMA - RESULTS FROM A SOCI□□INTERNATIONALE D□ONCOLOGIE P□DIATRIQUE EUROPE (SIOPE) CLINICAL TRIALS WORKSHOP. *Neuro-Oncology*, **2018**, 20, i143-i143	1	78
701	White Blood Cell BRCA1 Promoter Methylation Status and Ovarian Cancer Risk. *Annals of Internal Medicine*, **2018**, 168, 326-334	8	15
700	Exploring the prediction performance for breast cancer risk based on volumetric mammographic density at different thresholds. *Breast Cancer Research*, **2018**, 20, 49	8.3	6
699	Fanconi anemia with sun-sensitivity caused by a Xeroderma pigmentosum-associated missense mutation in XPF. *BMC Medical Genetics*, **2018**, 19, 7	2.1	7
698	Breast cancer risk in a screening cohort of Asian and white British/Irish women from Manchester UK. *BMC Public Health*, **2018**, 18, 178	4.1	9
697	Genetic variants of prospectively demonstrated phenocopies in kindreds. *Hereditary Cancer in Clinical Practice*, **2018**, 16, 4	2.3	6
696	A comparison of five methods of measuring mammographic density: a case-control study. *Breast Cancer Research*, **2018**, 20, 10	8.3	47
695	Psychological impact of providing women with personalised 10-year breast cancer risk estimates. *British Journal of Cancer*, **2018**, 118, 1648-1657	8.7	26
694	The prevalence of Lynch syndrome in women with endometrial cancer: a systematic review protocol. *Systematic Reviews*, **2018**, 7, 121	3	11
693	The prevention of breast cancer **2018**, 111-128		
692	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 3-18	11	27
691	Women's decision-making regarding risk-stratified breast cancer screening and prevention from the perspective of international healthcare professionals. *PLoS ONE*, **2018**, 13, e0197772	3.7	17
690	Schwannomatosis: a genetic and epidemiological study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1215-1219	5.5	55
689	C2 neurofibromas in neurofibromatosis type 1: genetic and imaging characteristics. *Journal of Neurosurgery: Spine*, **2018**, 30, 126-132	2.8	7
688	Risk-reducing mastectomy rates in the US: A closer examination of the Angelina Jolie effect.. *Journal of Clinical Oncology*, **2018**, 36, e13557-e13557	2.2	
687	Spinal ependymomas in NF2: a surgical disease?. *Journal of Neuro-Oncology*, **2018**, 136, 605-611	4.8	14

686	RAZOR: A Phase II Open Randomized Trial of Screening Plus Goserelin and Raloxifene Versus Screening Alone in Premenopausal Women at Increased Risk of Breast Cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2018**, 27, 58-66	4	2
685	Young age at first pregnancy does protect against early onset breast cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2018**, 167, 779-785	4.4	5
684	Low Lifetime Risk of Contralateral Breast Cancer in a Middle-Income Asian Country: Evidence to Guide Post-treatment Surveillance. *World Journal of Surgery*, **2018**, 42, 1270-1277	3.3	
683	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With or Mutations. *JNCI Cancer Spectrum*, **2018**, 2, pky078	4.6	10
682	Gene panel testing for breast cancer should not be used to confirm syndromic gene associations. *Npj Genomic Medicine*, **2018**, 3, 32	6.2	4
681	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. *JNCI Cancer Spectrum*, **2018**, 2, pky023	4.6	13
680	Exhaustive non-synonymous variants functionality prediction enables high resolution characterization of the neurofibromin architecture. *EBioMedicine*, **2018**, 36, 508-516	8.8	1
679	Risk-reducing mastectomy rates in the US: a closer examination of the Angelina Jolie effect. *Breast Cancer Research and Treatment*, **2018**, 171, 435-442	4.4	45
678	'For me it's about not feeling like I'm on a diet': a thematic analysis of women's experiences of an intermittent energy restricted diet to reduce breast cancer risk. *Journal of Human Nutrition and Dietetics*, **2018**, 31, 773-780	3.1	3
677	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
676	Pathological features and clinical behavior of Lynch syndrome-associated ovarian cancer. *Gynecologic Oncology*, **2017**, 144, 491-495	4.9	51
675	Baseline results from the UK SIGNIFY study: a whole-body MRI screening study in TP53 mutation carriers and matched controls. *Familial Cancer*, **2017**, 16, 433-440	3	39
674	Identifying High-Risk Women for Endometrial Cancer Prevention Strategies: Proposal of an Endometrial Cancer Risk Prediction Model. *Cancer Prevention Research*, **2017**, 10, 1-13	3.2	50
673	The BRCA1/2 Parent-of-Origin Effect on Breast Cancer Risk-Letter. *Cancer Epidemiology Biomarkers and Prevention*, **2017**, 26, 284	4	2
672	The impact of a panel of 18 SNPs on breast cancer risk in women attending a UK familial screening clinic: a case-control study. *Journal of Medical Genetics*, **2017**, 54, 111-113	5.8	42
671	Toxicity profile of bevacizumab in the UK Neurofibromatosis type 2 cohort. *Journal of Neuro-Oncology*, **2017**, 131, 117-124	4.8	32
670	Homozygous germ-line mutation of the PMS2 mismatch repair gene: a unique case report of constitutional mismatch repair deficiency (CMMRD). *BMC Medical Genetics*, **2017**, 18, 40	2.1	15
669	Pathology update to the Manchester Scoring System based on testing in over 4000 families. *Journal of Medical Genetics*, **2017**, 54, 674-681	5.8	37

668	Visual assessment of breast density using Visual Analogue Scales: observer variability, reader attributes and reading time **2017**,		3
667	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. *Clinical Cancer Research*, **2017**, 23, e46-e53	12.9	95
666	Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 2 and Related Disorders. *Clinical Cancer Research*, **2017**, 23, e54-e61	12.9	52
665	Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. *Clinical Cancer Research*, **2017**, 23, e62-e67	12.9	97
664	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
663	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. *Clinical Cancer Research*, **2017**, 23, e38-e45	12.9	245
662	Evaluation of a Stratified National Breast Screening Program in the United Kingdom: An Early Model-Based Cost-Effectiveness Analysis. *Value in Health*, **2017**, 20, 1100-1109	3.3	26
661	First evidence of genotype-phenotype correlations in Gorlin syndrome. *Journal of Medical Genetics*, **2017**, 54, 530-536	5.8	34
660	Cranial irradiation in childhood mimicking neurofibromatosis type II. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1635-1639	2.5	2
659	Neurofibromatosis type 2: Multiple intra-dermal tumors in a toddler. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1447-1449	2.5	1
658	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. *Gut*, **2017**, 66, 1657-1664	19.2	87
657	Urgent improvements needed to diagnose and manage Lynch syndrome. *BMJ, The*, **2017**, 356, j1388	5.9	9
656	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
655	Hearing optimisation in neurofibromatosis type 2: A systematic review. *Clinical Otolaryngology*, **2017**, 42, 1329-1337	1.8	30
654	The response of spinal cord ependymomas to bevacizumab in patients with neurofibromatosis Type 2. *Journal of Neurosurgery: Spine*, **2017**, 26, 474-482	2.8	21
653	False-negative MRI breast screening in high-risk women. *Clinical Radiology*, **2017**, 72, 207-216	2.9	12
652	Increased risk of breast cancer in neurofibromatosis type 1: current insights. *Breast Cancer: Targets and Therapy*, **2017**, 9, 531-536	3.9	19
651	Validation of a scale for assessing attitudes towards outcomes of genetic cancer testing among primary care providers and breast specialists. *PLoS ONE*, **2017**, 12, e0178447	3.7	4

650	Colorectal cancer incidence in carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. *Hereditary Cancer in Clinical Practice*, **2017**, 15, 18	2.3	27
649	A novel and fully automated mammographic texture analysis for risk prediction: results from two case-control studies. *Breast Cancer Research*, **2017**, 19, 114	8.3	25
648	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
647	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
646	Genetic Severity Score predicts clinical phenotype in NF2. *Journal of Medical Genetics*, **2017**, 54, 657-664	5.8	48
645	A randomised trial of screening with digital breast tomosynthesis plus conventional digital 2D mammography versus 2D mammography alone in younger higher risk women. *European Journal of Radiology*, **2017**, 94, 133-139	4.7	7
644	Association of Genetic Predisposition With Solitary Schwannoma or Meningioma in Children and Young Adults. *JAMA Neurology*, **2017**, 74, 1123-1129	17.2	36
643	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging: A Meta-analysis. *JAMA Oncology*, **2017**, 3, 1634-1639	13.4	107
642	Association of Mismatch Repair Mutation With Age at Cancer Onset in Lynch Syndrome: Implications for Stratified Surveillance Strategies. *JAMA Oncology*, **2017**, 3, 1702-1706	13.4	89
641	Preferences for breast cancer risk reduction among BRCA1/BRCA2 mutation carriers: a discrete-choice experiment. *Breast Cancer Research and Treatment*, **2017**, 165, 433-444	4.4	23
640	Risk algorithms that include pathology adjustment for HER2 amplification need to make further downward adjustments in likelihood scores. *Familial Cancer*, **2017**, 16, 173-179	3	2
639	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. *Clinical Cancer Research*, **2017**, 23, e107-e114	12.9	62
638	Cost-effectiveness of population based BRCA testing with varying Ashkenazi Jewish ancestry. *American Journal of Obstetrics and Gynecology*, **2017**, 217, 578.e1-578.e12	6.4	39
637	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. *Breast Cancer Research and Treatment*, **2017**, 161, 117-134	4.4	15
636	Revisiting neurofibromatosis type 2 diagnostic criteria to exclude LZTR1-related schwannomatosis. *Neurology*, **2017**, 88, 87-92	6.5	66
635	Creation of an international registry to support discovery in schwannomatosis. *American Journal of Medical Genetics, Part A*, **2017**, 173, 407-413	2.5	11
634	Bilateral Oophorectomy and Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	99
633	Body mass index and breast cancer survival: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2017**, 46, 1814-1822	7.8	27

632	BRCA and lynch syndrome-associated ovarian cancers behave differently. *Gynecologic Oncology Reports*, **2017**, 22, 108-109	1.3	7
631	Childhood neurofibromatosis type 2 (NF2) and related disorders: from bench to bedside and biologically targeted therapies. *Acta Otorhinolaryngologica Italica*, **2016**, 36, 345-367	2.8	24
630	Current status and recommendations for biomarkers and biobanking in neurofibromatosis. *Neurology*, **2016**, 87, S40-8	6.5	13
629	Current whole-body MRI applications in the neurofibromatoses: NF1, NF2, and schwannomatosis. *Neurology*, **2016**, 87, S31-9	6.5	49
628	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. *Breast Cancer Research*, **2016**, 18, 112	8.3	25
627	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
626	Outcomes of cochlear implantation in patients with neurofibromatosis type 2. *Cochlear Implants International*, **2016**, 17, 172-177	1.7	20
625	Auditory Brainstem Implantation in Neurofibromatosis Type 2: Experience From the Manchester Programme. *Otology and Neurotology*, **2016**, 37, 1267-74	2.6	20
624	Intermittent energy restriction induces changes in breast gene expression and systemic metabolism. *Breast Cancer Research*, **2016**, 18, 57	8.3	22
623	Sensitivity of BRCA1/2 testing in high-risk breast/ovarian/male breast cancer families: little contribution of comprehensive RNA/NGS panel testing. *European Journal of Human Genetics*, **2016**, 24, 1591-1597	5.3	21
622	Intensive breast screening in BRCA2 mutation carriers is associated with reduced breast cancer specific and all cause mortality. *Hereditary Cancer in Clinical Practice*, **2016**, 14, 8	2.3	25
621	Is there really an increased risk of early colorectal cancer in women with BRCA1 pathogenic mutations?. *Clinical Genetics*, **2016**, 89, 399	4	7
620	National survey of patients with Gorlin syndrome highlights poor awareness, multiple treatments and profound psychosocial impact of disease. *Journal of the European Academy of Dermatology and Venereology*, **2016**, 30, 371-3	4.6	2
619	Bevacizumab in neurofibromatosis type 2 (NF2) related vestibular schwannomas: a nationally coordinated approach to delivery and prospective evaluation. *Neuro-Oncology Practice*, **2016**, 3, 281-289	2.2	32
618	Vascular biomarkers derived from dynamic contrast-enhanced MRI predict response of vestibular schwannoma to antiangiogenic therapy in type 2 neurofibromatosis. *Neuro-Oncology*, **2016**, 18, 275-82	1	18
617	The Contribution of Whole Gene Deletions and Large Rearrangements to the Mutation Spectrum in Inherited Tumor Predisposing Syndromes. *Human Mutation*, **2016**, 37, 250-6	4.7	44
616	No strong evidence for increased risk of breast cancer 8-26 years after multiple mammograms in their 30s in females at moderate and high familial risk. *British Journal of Radiology*, **2016**, 89, 20150960	3.4	2
615	Genetic predisposition to cancer. *Medicine*, **2016**, 44, 65-68	0.6	1

614	Low prevalence of HER2 positivity amongst BRCA1 and BRCA2 mutation carriers and in primary BRCA screens. *Breast Cancer Research and Treatment*, **2016**, 155, 597-601	4.4	19
613	Genetic testing in a cohort of young patients with HER2-amplified breast cancer. *Annals of Oncology*, **2016**, 27, 467-73	10.3	16
612	Risk of contralateral breast cancer amongst BRCA1/2 mutation carriers. *Translational Cancer Research*, **2016**, 5, S1066-S1069	0.3	2
611	Improvement in risk prediction, early detection and prevention of breast cancer in the NHS Breast Screening Programme and family history clinics: a dual cohort study. *Programme Grants for Applied Research*, **2016**, 4, 1-210	1.5	48
610	Mammographic Density Over Time in Women With and Without Breast Cancer. *Lecture Notes in Computer Science*, **2016**, 291-298	0.9	1
609	Challenges and Opportunities in the Implementation of Risk-Based Screening for Breast Cancer **2016**, 165-187		
608	Should We Adjust Visually Assessed Mammographic Density for Observer Variability?. *Lecture Notes in Computer Science*, **2016**, 540-547	0.9	
607	Variations in Breast Density and Mammographic Risk Factors in Different Ethnic Groups. *Lecture Notes in Computer Science*, **2016**, 510-517	0.9	
606	Attitudes to contralateral risk reducing mastectomy among breast and plastic surgeons in England. *Annals of the Royal College of Surgeons of England*, **2016**, 98, 121-7	1.4	3
605	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
604	Finding Common Ground: Negotiating Across Cultures on Peace and Security Issues. *Global Policy*, **2016**, 7, 458-463	1.8	
603	Breast cancer risk feedback to women in the UK NHS breast screening population. *British Journal of Cancer*, **2016**, 114, 1045-52	8.7	48
602	Comprehensive RNA Analysis of the NF1 Gene in Classically Affected NF1 Affected Individuals Meeting NIH Criteria has High Sensitivity and Mutation Negative Testing is Reassuring in Isolated Cases With Pigmentary Features Only. *EBioMedicine*, **2016**, 7, 212-20	8.8	52
601	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. *American Journal of Human Genetics*, **2016**, 98, 857-868	11	14
600	A heritable form of SMARCE1-related meningiomas with important implications for follow-up and family screening. *Neurogenetics*, **2016**, 17, 83-9	3	34
599	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2016**, 18, 15	8.3	58
598	Biochemical Insights into Merlin/NF2 Pathophysiology and Biologically Targeted Therapies in Childhood NF2 and Related Forms. *Journal of Pediatric Biochemistry*, **2016**, 05, 120-130		
597	Beliefs about weight and breast cancer: an interview study with high risk women following a 12´month weight loss intervention. *Hereditary Cancer in Clinical Practice*, **2015**, 13, 1	2.3	21

596	Levels of soya aeroallergens during dockside unloading as measured by personal and static sampling. *Arhiv Za Higijenu Rada I Toksikologiju*, **2015**, 66, 23-9	1.7	2
595	Risk of contralateral breast cancer in BRCA1 and BRCA2 mutation carriers: a 30-year semi-prospective analysis. *Familial Cancer*, **2015**, 14, 531-8	3	34
594	Bilateral vestibular schwannomas in older patients: NF2 or chance?. *Journal of Medical Genetics*, **2015**, 52, 422-4	5.8	24
593	Lynch syndrome caused by MLH1 mutations is associated with an increased risk of breast cancer: a cohort study. *Journal of Medical Genetics*, **2015**, 52, 553-6	5.8	54
592	SMARCE1 mutations in pediatric clear cell meningioma: case report. *Journal of Neurosurgery: Pediatrics*, **2015**, 16, 296-300	2.1	23
591	Pathogenesis and management of type 2 neurofibromatosis. *Expert Opinion on Orphan Drugs*, **2015**, 3, 281-292	1.1	
590	Identification of novel genetic markers of breast cancer survival. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	38
589	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
588	General Practitioners and Breast Surgeons in France, Germany, Netherlands and the UK show variable breast cancer risk communication profiles. *BMC Cancer*, **2015**, 15, 243	4.8	6
587	Risk reducing salpingectomy and delayed oophorectomy in high risk women: views of cancer geneticists, genetic counsellors and gynaecological oncologists in the UK. *Familial Cancer*, **2015**, 14, 521-30	3	14
586	Obesity, Aspirin, and Risk of Colorectal Cancer in Carriers of Hereditary Colorectal Cancer: A Prospective Investigation in the CAPP2 Study. *Journal of Clinical Oncology*, **2015**, 33, 3591-7	2.2	71
585	Clinical and molecular predictors of mortality in neurofibromatosis 2: a UK national analysis of 1192 patients. *Journal of Medical Genetics*, **2015**, 52, 699-705	5.8	52
584	The Manchester guidelines for contralateral risk-reducing mastectomy. *World Journal of Surgical Oncology*, **2015**, 13, 237	3.4	21
583	A clinical and genetic analysis of multiple primary cancer referrals to genetics services. *European Journal of Human Genetics*, **2015**, 23, 581-7	5.3	12
582	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
581	Progress of hearing loss in neurofibromatosis type 2: implications for future management. *European Archives of Oto-Rhino-Laryngology*, **2015**, 272, 3143-50	3.5	10
580	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
579	Pediatric intracranial clear cell meningioma associated with a germline mutation of SMARCE1: a novel case. *Childs Nervous System*, **2015**, 31, 441-7	1.7	27

578	Osteoprotegerin (OPG), The Endogenous Inhibitor of Receptor Activator of NF-B Ligand (RANKL), is Dysregulated in BRCA Mutation Carriers. *EBioMedicine*, **2015**, 2, 1331-9	8.8	41
577	Can the breast screening appointment be used to provide risk assessment and prevention advice?. *Breast Cancer Research*, **2015**, 17, 84	8.3	27
576	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
575	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2015**, 17, 61	8.3	16
574	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
573	Longer term effects of the Angelina Jolie effect: increased risk-reducing mastectomy rates in BRCA carriers and other high-risk women. *Breast Cancer Research*, **2015**, 17, 143	8.3	64
572	Colonoscopy screening compliance and outcomes in patients with Lynch syndrome. *Colorectal Disease*, **2015**, 17, 38-46	2.1	39
571	Cognition in children with neurofibromatosis type 1: data from a population-based study. *Developmental Medicine and Child Neurology*, **2015**, 57, 645-651	3.3	22
570	In Silico Analysis of NF2 Gene Missense Mutations in Neurofibromatosis Type 2: From Genotype to Phenotype. *Otology and Neurotology*, **2015**, 36, 908-14	2.6	6
569	Contralateral risk-reducing mastectomy: review of risk factors and risk-reducing strategies. *International Journal of Surgical Oncology*, **2015**, 2015, 901046	0.9	13
568	Can diet and lifestyle prevent breast cancer: what is the evidence?. *American Society of Clinical Oncology Educational Book / ASCO American Society of Clinical Oncology Meeting*, **2015**, e66-73	7.1	52
567	Common variants modify the age of onset for basal cell carcinomas in Gorlin syndrome. *European Journal of Human Genetics*, **2015**, 23, 708-10	5.3	6
566	Mutations in LZTR1 add to the complex heterogeneity of schwannomatosis. *Neurology*, **2015**, 84, 141-7	6.5	67
565	Tumour characteristics and survival in familial breast cancer prospectively diagnosed by annual mammography. *Breast Cancer Research and Treatment*, **2015**, 152, 87-94	4.4	2
564	Autism spectrum disorder profile in neurofibromatosis type I. *Journal of Autism and Developmental Disorders*, **2015**, 45, 1649-57	4.6	42
563	The BRCA2 polymorphic stop codon: stuff or nonsense?. *Journal of Medical Genetics*, **2015**, 52, 642-5	5.8	8
562	Gene-panel sequencing and the prediction of breast-cancer risk. *New England Journal of Medicine*, **2015**, 372, 2243-57	59.2	587
561	Local mammographic density as a predictor of breast cancer **2015**,		2

560	Multiple synchronous sites of origin of vestibular schwannomas in neurofibromatosis Type 2. *Journal of Medical Genetics*, **2015**, 52, 557-62	5.8	29
559	Diagnosis, Management, and New Therapeutic Options in Childhood Neurofibromatosis Type 2 and Related Forms. *Seminars in Pediatric Neurology*, **2015**, 22, 240-58	2.9	58
558	Integrated smartcard solutions: do people want one card for all their services?. *Transportation Planning and Technology*, **2015**, 38, 534-551	1.6	4
557	Improving the uptake of predictive testing and colorectal screening in Lynch syndrome: a regional primary care survey. *Clinical Genetics*, **2015**, 87, 517-24	4	15
556	Rail grinding for the 21st century Taking a lead from the aerospace industry. *Proceedings of the Institution of Mechanical Engineers, Part F: Journal of Rail and Rapid Transit*, **2015**, 229, 457-465	1.4	18
555	Human in vivo and in vitro studies on gastrointestinal absorption of titanium dioxide nanoparticles. *Toxicology Letters*, **2015**, 233, 95-101	4.4	75
554	Breast surgeons' attitudes towards bilateral risk-reducing mastectomy: A National Survey of American Surgeons.. *Journal of Clinical Oncology*, **2015**, 33, 25-25	2.2	
553	Cancer risk and genotype-phenotype correlations in PTEN hamartoma tumor syndrome. *Familial Cancer*, **2014**, 13, 57-63	3	90
552	Mammographic surveillance in women aged 35-39 at enhanced familial risk of breast cancer (FH02). *Familial Cancer*, **2014**, 13, 13-21	3	11
551	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
550	MRI breast screening in high-risk women: cancer detection and survival analysis. *Breast Cancer Research and Treatment*, **2014**, 145, 663-72	4.4	105
549	Breast cancer risk assessment in 8,824 women attending a family history evaluation and screening programme. *Familial Cancer*, **2014**, 13, 189-96	3	19
548	Fanconi anaemia, BRCA2 mutations and childhood cancer: a developmental perspective from clinical and epidemiological observations with implications for genetic counselling. *Journal of Medical Genetics*, **2014**, 51, 71-5	5.8	38
547	A pooled analysis of the outcome of prospective colonoscopic surveillance for familial colorectal cancer. *International Journal of Cancer*, **2014**, 134, 939-47	7.5	20
546	Nuclear Deterrence in Asia and the Pacific. *Asia and the Pacific Policy Studies*, **2014**, 1, 91-111	2.3	4
545	Germline SMARCE1 mutations predispose to both spinal and cranial clear cell meningiomas. *Journal of Pathology*, **2014**, 234, 436-40	9.4	81
544	Tumour MLH1 promoter region methylation testing is an effective prescreen for Lynch Syndrome (HNPCC). *Journal of Medical Genetics*, **2014**, 51, 789-96	5.8	50
543	Update from the 2013 International Neurofibromatosis Conference. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2969-78	2.5	11

542	Long-term prospective clinical follow-up after BRCA1/2 presymptomatic testing: BRCA2 risks higher than in adjusted retrospective studies. *Journal of Medical Genetics*, **2014**, 51, 573-80	5.8	14
541	Management of women at high risk of breast cancer. *BMJ, The*, **2014**, 348, g2756	5.9	21
540	Risk determination and prevention of breast cancer. *Breast Cancer Research*, **2014**, 16, 446	8.3	180
539	Longitudinal evaluation of quality of life in 288 patients with neurofibromatosis 2. *Journal of Neurology*, **2014**, 261, 963-9	5.5	30
538	Intronic splicing mutations in PTCH1 cause Gorlin syndrome. *Familial Cancer*, **2014**, 13, 477-80	3	22
537	SMARCB1 mutations in schwannomatosis and genotype correlations with rhabdoid tumors. *Cancer Genetics*, **2014**, 207, 373-8	2.3	56
536	Threshold for genetic testing in women with breast cancer needs to be determined. *BMJ, The*, **2014** , 348, g1863	5.9	1
535	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
534	BCRT response to Moller. *Breast Cancer Research and Treatment*, **2014**, 148, 693	4.4	1
533	Can multiple SNP testing in BRCA2 and BRCA1 female carriers be used to improve risk prediction models in conjunction with clinical assessment?. *BMC Medical Informatics and Decision Making*, **2014** , 14, 87	3.6	8
532	Clinical response to bevacizumab in schwannomatosis. *Neurology*, **2014**, 83, 1986-7	6.5	26
531	Uptake of tamoxifen in consecutive premenopausal women under surveillance in a high-risk breast cancer clinic. *British Journal of Cancer*, **2014**, 110, 1681-7	8.7	67
530	Distribution of breast cancer risk from SNPs and classical risk factors in women of routine screening age in the UK. *British Journal of Cancer*, **2014**, 110, 827-8	8.7	27
529	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
528	Review of radiation therapy services for neurofibromatosis (NF2) patients in England. *British Journal of Neurosurgery*, **2014**, 28, 16-9	1	7
527	Current Concepts in Management of Vestibular Schwannomas in Neurofibromatosis Type 2. *Current Otorhinolaryngology Reports*, **2014**, 2, 248-255	0.5	
526	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
525	The Fragile X Protein binds mRNA s involved in cancer progression and modulates metastasis formation. *EMBO Molecular Medicine*, **2014**, 6, 567-568	12	78

524	Response to Santoro et al. *Breast Cancer Research and Treatment*, **2014**, 147, 689	4.4	
523	Use of risk-reducing surgeries in a prospective cohort of 1,499 BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2014**, 148, 397-406	4.4	42
522	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
521	Breast cancer risk in young women in the national breast screening programme: implications for applying NICE guidelines for additional screening and chemoprevention. *Cancer Prevention Research*, **2014**, 7, 993-1001	3.2	23
520	Ipsilateral cochlear implantation after cochlear nerve preserving vestibular schwannoma surgery in patients with neurofibromatosis type 2. *Otology and Neurotology*, **2014**, 35, 43-51	2.6	52
519	The Genetics of Vestibular Schwannoma. *Current Otorhinolaryngology Reports*, **2014**, 2, 226-234	0.5	7
518	Lymphocyte telomere length is long in BRCA1 and BRCA2 mutation carriers regardless of cancer-affected status. *Cancer Epidemiology Biomarkers and Prevention*, **2014**, 23, 1018-24	4	12
517	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2014**, 16, 3416	8.3	46
516	Cancer risk communication, predictive testing and management in France, Germany, the Netherlands and the UK: general practitioners' and breast surgeons' current practice and preferred practice responsibilities. *Journal of Community Genetics*, **2014**, 5, 69-79	2.5	12
515	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget*, **2014**, 5, 8223-34	3.3	21
514	Small Bowel Cancer in the UK. *American Journal of Gastroenterology*, **2014**, 109, S116-S117	0.7	
513	Cranial Meningioma in Neurofibromatosis Type 2 Patients: Role of Mutations. *Tumors of the Central Nervous System*, **2014**, 271-276		
512	The genetics of breast cancer, risk-reducing surgery and prevention **2014**, 127-145		
511	Contralateral breast cancer risk in BRCA1/2-positive families needs to be adjusted for phenocopy rates particularly in second-degree untested relatives. *Breast Cancer Research*, **2013**, 15, 401	8.3	1
510	Neurofibromatosis type 2 (NF2): diagnosis and management. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 115, 957-67	3	45
509	Contralateral mastectomy improves survival in women with BRCA1/2-associated breast cancer. *Breast Cancer Research and Treatment*, **2013**, 140, 135-42	4.4	119
508	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. *Nature Genetics*, **2013**, 45, 295-8	36.3	162
507	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

506	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
505	Evaluation of SDHB, SDHD and VHL gene susceptibility testing in the assessment of individuals with non-syndromic phaeochromocytoma, paraganglioma and head and neck paraganglioma. *Clinical Endocrinology*, **2013**, 78, 898-906	3.4	48
504	Systematic review of the impact of registration and screening on colorectal cancer incidence and mortality in familial adenomatous polyposis and Lynch syndrome. *British Journal of Surgery*, **2013**, 100, 1719-31	5.3	84
503	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
502	Critical research gaps and translational priorities for the successful prevention and treatment of breast cancer. *Breast Cancer Research*, **2013**, 15, R92	8.3	248
501	Risk-reducing surgery increases survival in BRCA1/2 mutation carriers unaffected at time of family referral. *Breast Cancer Research and Treatment*, **2013**, 142, 611-8	4.4	45
500	Outcome of translabyrinthine surgery for vestibular schwannoma in neurofibromatosis type 2. *British Journal of Neurosurgery*, **2013**, 27, 446-53	1	21
499	The MSH2 c.388_389del mutation shows a founder effect in Portuguese Lynch syndrome families. *Clinical Genetics*, **2013**, 84, 244-50	4	11
498	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
497	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. *Nature*, **2013**, 493, 406-10	50.4	191
496	Neurofibromatosis type 1 and autism spectrum disorder. *Pediatrics*, **2013**, 132, e1642-8	7.4	116
495	International variation in physicians' attitudes towards prophylactic mastectomy - comparison between France, Germany, the Netherlands and the United Kingdom. *European Journal of Cancer*, **2013**, 49, 2798-805	7.5	20
494	Breast cancer prevention: SERMs come of age. *Lancet, The*, **2013**, 381, 1795-7	40	4
493	Contralateral risk reducing mastectomy A national survey of surgeons' practices and perceptions. *European Journal of Surgical Oncology*, **2013**, 39, S64	3.6	4
492	Mutation type and position varies between mosaic and inherited NF2 and correlates with disease severity. *Clinical Genetics*, **2013**, 83, 594-5	4	21
491	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
490	Haplotype analysis of the 185delAG BRCA1 mutation in ethnically diverse populations. *European Journal of Human Genetics*, **2013**, 21, 212-6	5.3	32
489	Is multiple SNP testing in BRCA2 and BRCA1 female carriers ready for use in clinical practice? Results from a large Genetic Centre in the UK. *Clinical Genetics*, **2013**, 84, 37-42	4	11

488	Cancer risk in Lynch Syndrome. *Familial Cancer*, **2013**, 12, 229-40	3	138
487	Familial breast cancer: summary of updated NICE guidance. *BMJ, The*, **2013**, 346, f3829	5.9	48
486	The spectrum of urological malignancy in Lynch syndrome. *Familial Cancer*, **2013**, 12, 57-63	3	39
485	Correspondence: Humanitarian Intervention and the Responsibility to Protect. *International Security*, **2013**, 37, 199-214	4.2	24
484	Reproductive decision-making in young female carriers of a BRCA mutation. *Human Reproduction*, **2013**, 28, 1006-12	5.7	46
483	Can the diagnosis of NF1 be excluded clinically? A lack of pigmentary findings in families with spinal neurofibromatosis demonstrates a limitation of clinical diagnosis. *Journal of Medical Genetics*, **2013**, 50, 606-13	5.8	23
482	Increased rate of missense/in-frame mutations in individuals with NF1-related pulmonary stenosis: a novel genotype-phenotype correlation. *European Journal of Human Genetics*, **2013**, 21, 535-9	5.3	24
481	Increased rate of phenocopies in all age groups in BRCA1/BRCA2 mutation kindred, but increased prospective breast cancer risk is confined to BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2013**, 22, 2269-76	4	12
480	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
479	Recommendations for imaging tumor response in neurofibromatosis clinical trials. *Neurology*, **2013**, 81, S33-40	6.5	81
478	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
477	Can manipulation of splicing offer gene therapy possibilities to those with tumour-prone disorders?. *European Journal of Human Genetics*, **2013**, 21, 701-2	5.3	1
476	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
475	Hearing and facial function outcomes for neurofibromatosis 2 clinical trials. *Neurology*, **2013**, 81, S25-32	6.5	27
474	Metachronous colorectal cancer risk in patients with a moderate family history. *Colorectal Disease*, **2013**, 15, 309-16	2.1	6
473	Successful radiofrequency ablation of an anterior abdominal wall desmoid in familial adenomatous polyposis. *Colorectal Disease*, **2013**, 15, e160-3	2.1	7
472	Ovarian cancer among 8,005 women from a breast cancer family history clinic: no increased risk of invasive ovarian cancer in families testing negative for BRCA1 and BRCA2. *Journal of Medical Genetics*, **2013**, 50, 368-72	5.8	21
471	Optimal age to start preventive measures in women with BRCA1/2 mutations or high familial breast cancer risk. *International Journal of Cancer*, **2013**, 133, 156-63	7.5	15

470	The fragile X protein binds mRNAs involved in cancer progression and modulates metastasis formation. *EMBO Molecular Medicine*, **2013**, 5, 1523-36	12	78
469	Prophylactic mastectomy and breast cancer-- reply. *British Journal of Hospital Medicine (London, England: 2005)*, **2013**, 74, 595	0.8	
468	Same task, same observers, different values: the problem with visual assessment of breast density **2013**,		2
467	Key genetic considerations in the management of suspected hereditary colorectal cancer. *Colorectal Cancer*, **2013**, 2, 31-41	0.8	
466	RIC-3 differentially modulates α2 and β4 nicotinic receptor assembly, expression, and nicotine-induced receptor upregulation. *BMC Neuroscience*, **2013**, 14, 47	3.2	20
465	English consensus protocol evaluating candidacy for auditory brainstem and cochlear implantation in neurofibromatosis type 2. *Otology and Neurotology*, **2013**, 34, 1743-7	2.6	20
464	Role of Engrailed-2 (EN2) as a prostate cancer detection biomarker in genetically high risk men. *Scientific Reports*, **2013**, 3, 2059	4.9	18
463	Reduced life expectancy seen in hereditary diseases which predispose to early-onset tumors. *The Application of Clinical Genetics*, **2013**, 6, 53-61	3.1	13
462	Final results of 4-monthly screening in the UK Familial Ovarian Cancer Screening Study (UKFOCSS Phase 2).. *Journal of Clinical Oncology*, **2013**, 31, 5507-5507	2.2	5
461	Evaluation of mammographic surveillance services in women aged 40-49 years with a moderate family history of breast cancer: a single-arm cohort study. *Health Technology Assessment*, **2013**, 17, vii-xiv, 1-95	4.4	8
460	Genetic predisposition to cancer. *Medicine*, **2012**, 40, 29-33	0.6	2
459	Genotype-phenotype correlation in colorectal polyposis. *Clinical Genetics*, **2012**, 81, 521-31	4	37
458	Familial breast cancer. *Clinical Genetics*, **2012**, 82, 105-14	4	122
457	Clinical presentation, immunohistochemistry and electron microscopy indicate neurofibromatosis type 2-associated gliomas to be spinal ependymomas. *Neuropathology*, **2012**, 32, 611-6	2	24
456	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
455	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
454	Consensus recommendations for current treatments and accelerating clinical trials for patients with neurofibromatosis type 2. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 24-41	2.5	88
453	Vestibular schwannomas occur in schwannomatosis and should not be considered an exclusion criterion for clinical diagnosis. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 215-9	2.5	41

452	Genetic predisposition to peripheral nerve neoplasia: diagnostic criteria and pathogenesis of neurofibromatoses, Carney complex, and related syndromes. *Acta Neuropathologica*, **2012**, 123, 349-67	14.3	58
451	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
450	Genetic testing and screening of individuals at risk of NF2. *Clinical Genetics*, **2012**, 82, 416-24	4	41
449	Breast and ovarian cancer risk and risk reduction in Jewish BRCA1/2 mutation carriers. *Journal of Clinical Oncology*, **2012**, 30, 1321-8	2.2	25
448	Prevention of breast cancer in the context of a national breast screening programme. *Journal of Internal Medicine*, **2012**, 271, 321-30	10.8	24
447	Effects of BRCA2 cis-regulation in normal breast and cancer risk amongst BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R63	8.3	15
446	Assessing individual breast cancer risk within the U.K. National Health Service Breast Screening Program: a new paradigm for cancer prevention. *Cancer Prevention Research*, **2012**, 5, 943-51	3.2	79
445	Endometrial cancer and venous thromboembolism in women under age 50 who take tamoxifen for prevention of breast cancer: a systematic review. *Cancer Treatment Reviews*, **2012**, 38, 318-28	14.4	67
444	Malignant peripheral nerve sheath tumours in inherited disease. *Clinical Sarcoma Research*, **2012**, 2, 17	2.5	76
443	Developing national guidance on genetic testing for breast cancer predisposition: the role of economic evidence?. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 580-91	1.6	8
442	BRCA carriers, prophylactic salpingo-oophorectomy and menopause: clinical management considerations and recommendations. *Women's Health*, **2012**, 8, 543-55	3	61
441	Long-term effect of resistant starch on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. *Lancet Oncology, The*, **2012**, 13, 1242-9	21.7	70
440	Loss of SUFU function in familial multiple meningioma. *American Journal of Human Genetics*, **2012**, 91, 520-6	11	103
439	Breast cancer risk and 6q22.33: combined results from Breast Cancer Association Consortium and Consortium of Investigators on Modifiers of BRCA1/2. *PLoS ONE*, **2012**, 7, e35706	3.7	10
438	Life expectancy in hereditary cancer predisposing diseases: an observational study. *Journal of Medical Genetics*, **2012**, 49, 264-9	5.8	98
437	An improved coverage and spatial resolution--using dual injection dynamic contrast-enhanced (ICE-DICE) MRI: a novel dynamic contrast-enhanced technique for cerebral tumors. *Magnetic Resonance in Medicine*, **2012**, 68, 452-62	4.4	21
436	Surveillance of women at increased risk of breast cancer using mammography and clinical breast examination: further evidence of benefit. *International Journal of Cancer*, **2012**, 131, 417-25	7.5	22
435	Association of PHB 1630 C>T and MTHFR 677 C>T polymorphisms with breast and ovarian cancer risk in BRCA1/2 mutation carriers: results from a multicenter study. *British Journal of Cancer*, **2012**, 106, 2016-24	8.7	25

434	Gene-gene interactions in breast cancer susceptibility. *Human Molecular Genetics*, **2012**, 21, 958-62	5.6	33
433	High sensitivity for BRCA1/2 mutations in breast/ovarian kindreds: are there still other breast/ovary genes to be discovered?. *Breast Cancer Research and Treatment*, **2012**, 134, 895-7	4.4	4
432	Risk of cancer other than breast or ovarian in individuals with BRCA1 and BRCA2 mutations. *Familial Cancer*, **2012**, 11, 235-42	3	188
431	Comment on the article "Germline SMARCB1 mutation predisposes to multiple meningiomas and schwannomas with preferential location of cranial meningiomas at the falx cerebri" by van den Munckhof et al. *Neurogenetics*, **2012**, 13, 103-4	3	2
430	Frequency of SMARCB1 mutations in familial and sporadic schwannomatosis. *Neurogenetics*, **2012**, 13, 141-5	3	86
429	Lack of caveolin-1 (P132L) somatic mutations in breast cancer. *Breast Cancer Research and Treatment*, **2012**, 132, 1185-6	4.4	5
428	Polymorphisms of CYP19A1 and response to aromatase inhibitors in metastatic breast cancer patients. *Breast Cancer Research and Treatment*, **2012**, 133, 1191-8	4.4	32
427	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1362-70	4	20
426	Attitudes to reproductive genetic testing in women who had a positive BRCA test before having children: a qualitative analysis. *European Journal of Human Genetics*, **2012**, 20, 4-10	5.3	55
425	Are we ready for online tools in decision making for BRCA1/2 mutation carriers?. *Journal of Clinical Oncology*, **2012**, 30, 471-3	2.2	8
424	Detection and management of women at increased risk of breast cancer. *Clinical Practice (London, England)*, **2012**, 9, 391-401	3	
423	Two out of three required: a case of incomplete Carney triad. *International Journal of Surgical Pathology*, **2012**, 20, 265-8	1.2	2
422	Fine-mapping CASP8 risk variants in breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 176-81	4	19
421	BRCA1 testing should be offered to individuals with triple-negative breast cancer diagnosed below 50 years. *British Journal of Cancer*, **2012**, 106, 1234-8	8.7	76
420	Uptake of risk-reducing salpingo-oophorectomy in women carrying a BRCA1 or BRCA2 mutation: evidence for lower uptake in women affected by breast cancer and older women. *British Journal of Cancer*, **2012**, 106, 775-9	8.7	18
419	Germline RAD51C mutations confer susceptibility to ovarian cancer. *Nature Genetics*, **2012**, 44, 475-6; author reply 476	36.3	190
418	Breast cancer risk for noncarriers of family-specific BRCA1 and BRCA2 mutations: more trouble with phenocopies. *Journal of Clinical Oncology*, **2012**, 30, 1142-3; author reply 1143-4	2.2	5
417	Differences in natural history between breast cancers in BRCA1 and BRCA2 mutation carriers and effects of MRI screening-MRISC, MARIBS, and Canadian studies combined. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1458-68	4	72

416	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 645-57	4	44
415	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
414	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
413	Volumetric and Area-Based Breast Density Measurement in the Predicting Risk of Cancer at Screening (PROCAS) Study. *Lecture Notes in Computer Science*, **2012**, 228-235	0.9	6
412	Volumetric and Area-Based Measures of Mammographic Density in Women with and without Cancer. *Lecture Notes in Computer Science*, **2012**, 589-595	0.9	3
411	Ethnic Variation in Volumetric Breast Density. *Lecture Notes in Computer Science*, **2012**, 127-133	0.9	1
410	Empirical development of improved diagnostic criteria for neurofibromatosis 2. *Genetics in Medicine*, **2011**, 13, 576-81	8.1	67
409	Neurofibromatosis type 2. *Advances in Oto-Rhino-Laryngology*, **2011**, 70, 91-98	1.7	25
408	A comparative study of quantitative immunohistochemistry and quantum dot immunohistochemistry for mutation carrier identification in Lynch syndrome. *Journal of Clinical Pathology*, **2011**, 64, 208-14	3.9	13
407	Neurofibromatoses in Clinical Practice **2011**,		7
406	Modification of BRCA1-Associated Breast and Ovarian Cancer Risk by BRCA1-Interacting Genes. *Cancer Research*, **2011**, 71, 5792-805	10.1	44
405	Malignant peripheral nerve sheath tumours in NF1: improved survival in women and in recent years. *European Journal of Cancer*, **2011**, 47, 2723-8	7.5	29
404	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
403	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2011**, 13, R110	8.3	62
402	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
401	Hyperplastic polyps are innocuous lesions in hereditary nonpolyposis colorectal cancers. *International Journal of Surgical Oncology*, **2011**, 2011, 653163	0.9	2
400	Targeted prostate cancer screening in men with mutations in BRCA1 and BRCA2 detects aggressive prostate cancer: preliminary analysis of the results of the IMPACT study. *BJU International*, **2011**, 107, 28-39	5.6	76
399	Psychological impact and acceptability of magnetic resonance imaging and X-ray mammography: the MARIBS Study. *British Journal of Cancer*, **2011**, 104, 578-86	8.7	18

398	Evaluation of the XRCC1 gene as a phenotypic modifier in BRCA1/2 mutation carriers. Results from the consortium of investigators of modifiers of BRCA1/BRCA2. *British Journal of Cancer*, **2011**, 104, 1356-61	8.7	6
397	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
396	Neurofibromatosis Type 2 (NF2) **2011**, 47-70		3
395	Familial breast cancer: is it time to move from a reactive to a proactive role?. *Familial Cancer*, **2011**, 10, 501-3	3	8
394	Comprehensive CYP2D6 genotype and adherence affect outcome in breast cancer patients treated with tamoxifen monotherapy. *Breast Cancer Research and Treatment*, **2011**, 125, 279-87	4.4	76
393	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
392	Genetic testing for familial/hereditary breast cancer-comparison of guidelines and recommendations from the UK, France, the Netherlands and Germany. *Journal of Community Genetics*, **2011**, 2, 53-69	2.5	56
391	Hormone replacement therapy and breast cancer. *Recent Results in Cancer Research*, **2011**, 188, 115-24	1.5	19
390	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
389	Prevalence of BRCA1 and BRCA2 mutations in triple negative breast cancer. *Journal of Medical Genetics*, **2011**, 48, 520-2	5.8	59
388	A rapid agonist application system for fast activation of ligand-gated ion channels. *Journal of Neuroscience Methods*, **2011**, 198, 246-54	3	8
387	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
386	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 3304-21	5.6	62
385	Common genetic variation at BARD1 is not associated with breast cancer risk in BRCA1 or BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 1032-8	4	13
384	A molecular analysis of individuals with neurofibromatosis type 1 (NF1) and optic pathway gliomas (OPGs), and an assessment of genotype-phenotype correlations. *Journal of Medical Genetics*, **2011**, 48, 256-60	5.8	74
383	Frequent hSNF5/INI1 germline mutations in patients with rhabdoid tumor. *Clinical Cancer Research*, **2011**, 17, 31-8	12.9	153
382	Cranial meningiomas in 411 neurofibromatosis type 2 (NF2) patients with proven gene mutations: clear positional effect of mutations, but absence of female severity effect on age at onset. *Journal of Medical Genetics*, **2011**, 48, 261-5	5.8	72
381	Basal cell carcinomas in gorlin syndrome: a review of 202 patients. *Journal of Skin Cancer*, **2011**, 2011, 217378	1.4	70

380	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
379	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
378	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
377	Ethnopolitical Conflict: When is it Right to Intervene?. *Ethnopolitics*, **2011**, 10, 115-123	0.7	2
376	Germline mutations in RAD51D confer susceptibility to ovarian cancer. *Nature Genetics*, **2011**, 43, 879-882	36.3	379
375	Menopausal symptoms and bone health in women undertaking risk reducing bilateral salpingo-oophorectomy: significant bone health issues in those not taking HRT. *British Journal of Cancer*, **2011**, 105, 22-7	8.7	46
374	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
373	Is hormone replacement therapy (HRT) following risk-reducing salpingo-oophorectomy (RRSO) in BRCA1 (B1)- and BRCA2 (B2)-mutation carriers associated with an increased risk of breast cancer?. *Journal of Clinical Oncology*, **2011**, 29, 1501-1501	2.2	13
372	The relationship between patients' perception of the effects of neurofibromatosis type 2 and the domains of the Short Form-36. *Clinical Otolaryngology*, **2010**, 35, 291-9	1.8	21
371	Rates of loss of heterozygosity and mitotic recombination in NF2 schwannomas, sporadic vestibular schwannomas and schwannomatosis schwannomas. *Oncogene*, **2010**, 29, 6216-21	9.2	77
370	Long-term outcomes of breast cancer in women aged 30 years or younger, based on family history, pathology and BRCA1/BRCA2/TP53 status. *British Journal of Cancer*, **2010**, 102, 1091-8	8.7	40
369	Prostate cancer in BRCA2 germline mutation carriers is associated with poorer prognosis. *British Journal of Cancer*, **2010**, 103, 918-24	8.7	109
368	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
367	Semiquantitative assessment of immunohistochemistry for mismatch repair proteins in Lynch syndrome. *Histopathology*, **2010**, 56, 331-44	7.3	24
366	Genome-wide association study identifies five new breast cancer susceptibility loci. *Nature Genetics* , **2010**, 42, 504-7	36.3	582
365	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
364	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
363	Further genotype--phenotype correlations in neurofibromatosis 2. *Clinical Genetics*, **2010**, 77, 163-70	4	73

362	The rs10993994 risk allele for prostate cancer results in clinically relevant changes in microseminoprotein-beta expression in tissue and urine. *PLoS ONE*, **2010**, 5, e13363	3.7	68
361	SMARCB1 mutations are not a common cause of multiple meningiomas. *Journal of Medical Genetics*, **2010**, 47, 567-8	5.8	38
360	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
359	Risk of breast cancer in male BRCA2 carriers. *Journal of Medical Genetics*, **2010**, 47, 710-1	5.8	74
358	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
357	Use of a closed set questionnaire to measure primary and secondary effects of neurofibromatosis type 2. *Journal of Laryngology and Otology*, **2010**, 124, 720-8	1.8	37
356	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
355	A novel HER2-positive breast cancer phenotype arising from germline TP53 mutations. *Journal of Medical Genetics*, **2010**, 47, 771-4	5.8	85
354	Association of the variants CASP8 D302H and CASP10 V410I with breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 2859-68	4	32
353	The impact of screening and genetic registration on mortality and colorectal cancer incidence in familial adenomatous polyposis. *Gut*, **2010**, 59, 1378-82	19.2	43
352	Survival in women with MMR mutations and ovarian cancer: a multicentre study in Lynch syndrome kindreds. *Journal of Medical Genetics*, **2010**, 47, 99-102	5.8	53
351	BRCA1, BRCA2 and CHEK2 c.1100 delC mutations in patients with double primaries of the breasts and/or ovaries. *Journal of Medical Genetics*, **2010**, 47, 561-6	5.8	7
350	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
349	Uptake of breast cancer prevention and screening trials. *Journal of Medical Genetics*, **2010**, 47, 853-5	5.8	15
348	Evidence for SMAD3 as a modifier of breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2010**, 12, R102	8.3	21
347	Breast cancer susceptibility variants alter risks in familial disease. *Journal of Medical Genetics*, **2010**, 47, 126-31	5.8	34
346	Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups (update from 2002). *Gut*, **2010**, 59, 666-89	19.2	843
345	Development of a scoring system to screen for BRCA1/2 mutations. *Methods in Molecular Biology*, **2010**, 653, 237-47	1.4	6

344	Vestibular schwannoma: role of conservative management. *Journal of Laryngology and Otology*, **2010**, 124, 251-7	1.8	42
343	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
342	Occult ovarian cancers identified at risk-reducing salpingo-oophorectomy in a prospective cohort of BRCA1/2 mutation carriers. *Breast Cancer Research and Treatment*, **2010**, 124, 195-203	4.4	44
341	Mutation and association analysis of GEN1 in breast cancer susceptibility. *Breast Cancer Research and Treatment*, **2010**, 124, 283-8	4.4	7
340	Childhood predictive genetic testing for Li-Fraumeni syndrome. *Familial Cancer*, **2010**, 9, 65-9	3	20
339	RASSF1A polymorphism in familial breast cancer. *Familial Cancer*, **2010**, 9, 263-5	3	15
338	Breast cancer susceptibility variants alter risk in familial ovarian cancer. *Familial Cancer*, **2010**, 9, 503-6	3	4
337	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
336	Isolated unilateral vestibular schwannomas do not harbor HRAS mutations. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1586-7	2.5	
335	Cancer occurrence during follow-up of the CAPP2 study -aspirin use for up to four years significantly reduces Lynch syndrome cancers for up to several years after completion of therapy. *Hereditary Cancer in Clinical Practice*, **2010**, 8, O5	2.3	1
334	Update on genetic predisposition to breast cancer. *Expert Review of Anticancer Therapy*, **2009**, 9, 1103-13	3.5	10
333	An update on age related mosaic and offspring risk in neurofibromatosis 2 (NF2). *Journal of Medical Genetics*, **2009**, 46, 792	5.8	18
332	Eligibility for magnetic resonance imaging screening in the United Kingdom: effect of strict selection criteria and anonymous DNA testing on breast cancer incidence in the MARIBS Study. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 2123-31	4	13
331	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
330	Cancers in BRCA1 and BRCA2 carriers and in women at high risk for breast cancer: MR imaging and mammographic features. *Radiology*, **2009**, 252, 358-68	20.5	58
329	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
328	Outcome from surgery for vestibular schwannomas in children. *British Journal of Neurosurgery*, **2009**, 23, 226-31	1	18
327	Addition of pathology and biomarker information significantly improves the performance of the Manchester scoring system for BRCA1 and BRCA2 testing. *Journal of Medical Genetics*, **2009**, 46, 811-7	5.8	65

326	Consensus recommendations to accelerate clinical trials for neurofibromatosis type 2. *Clinical Cancer Research*, **2009**, 15, 5032-5039	12.9	61
325	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
324	Sensitization to wheat flour and enzymes and associated respiratory symptoms in British bakers. *American Journal of Industrial Medicine*, **2009**, 52, 133-40	2.7	20
323	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. *Breast Cancer Research and Treatment*, **2009**, 117, 371-9	4.4	10
322	Strategies for endometrial screening in the Lynch syndrome population: a patient acceptability study. *Familial Cancer*, **2009**, 8, 431-9	3	11
321	Pathogenesis of vestibular schwannoma in ring chromosome 22. *BMC Medical Genetics*, **2009**, 10, 97	2.1	12
320	Risk-reducing surgery for ovarian cancer: outcomes in 300 surgeries suggest a low peritoneal primary risk. *European Journal of Human Genetics*, **2009**, 17, 1381-5	5.3	26
319	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393
318	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2009**, 101, 1456-60	8.7	17
317	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). *British Journal of Cancer*, **2009**, 101, 2048-54	8.7	13
316	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
315	Comparison of proactive and usual approaches to offering predictive testing for BRCA1/2 mutations in unaffected relatives. *Clinical Genetics*, **2009**, 75, 124-32	4	33
314	Increased colorectal cancer incidence in obligate carriers of heterozygous mutations in MUTYH. *Gastroenterology*, **2009**, 137, 489-94, 494.e1; quiz 725-6	13.3	90
313	Neurofibromatosis type 2 (NF2): a clinical and molecular review. *Orphanet Journal of Rare Diseases*, **2009**, 4, 16	4.2	294
312	Neurofibromatosis 2 [Bilateral acoustic neurofibromatosis, central neurofibromatosis, NF2, neurofibromatosis type II]. *Genetics in Medicine*, **2009**, 11, 599-610	8.1	98
311	Assessing the usefulness of a novel MRI-based breast density estimation algorithm in a cohort of women at high genetic risk of breast cancer: the UK MARIBS study. *Breast Cancer Research*, **2009**, 11, R80	8.3	70
310	Risk reducing mastectomy: outcomes in 10 European centres. *Journal of Medical Genetics*, **2009**, 46, 254-8	5.8	73
309	Screening for familial ovarian cancer: poor survival of BRCA1/2 related cancers. *Journal of Medical Genetics*, **2009**, 46, 593-7	5.8	103

308	Deciphering the genetics of hereditary non-syndromic colorectal cancer. *European Journal of Human Genetics*, **2008**, 16, 1477-86	5.3	29
307	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
306	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
305	Probability of BRCA1/2 mutation varies with ovarian histology: results from screening 442 ovarian cancer families. *Clinical Genetics*, **2008**, 73, 338-45	4	34
304	Colorectal cancer in HNPCC: cumulative lifetime incidence, survival and tumour distribution. A report of 121 families with proven mutations. *Clinical Genetics*, **2008**, 74, 233-42	4	75
303	Diagnosed with breast cancer while on a family history screening programme: an exploratory qualitative study. *European Journal of Cancer Care*, **2008**, 17, 245-52	2.4	12
302	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
301	A computer-based aid for communication between patients with limited English and their clinicians, using symbols and digitised speech. *International Journal of Medical Informatics*, **2008**, 77, 507-17	5.3	6
300	Genetic predisposition to cancer. *Medicine*, **2008**, 36, 50-54	0.6	
299	Trigeminal schwannomas. *British Journal of Neurosurgery*, **2008**, 22, 729-38	1	45
298	Assessment of in vitro sperm characteristics in relation to fertility in dairy bulls. *Animal Reproduction Science*, **2008**, 103, 201-14	2.1	104
297	Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer. *Human Molecular Genetics*, **2008**, 17, 3720-7	5.6	57
296	Red clover isoflavones are safe and well tolerated in women with a family history of breast cancer. *Menopause International*, **2008**, 14, 6-12		47
295	Prevalence of adenomas and hyperplastic polyps in mismatch repair mutation carriers among CAPP2 participants: report by the colorectal adenoma/carcinoma prevention programme 2. *Journal of Clinical Oncology*, **2008**, 26, 3434-9	2.2	29
294	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
293	Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. *Journal of Medical Genetics*, **2008**, 45, 332-9	5.8	141
292	A pilot study of compositional analysis of the breast and estimation of breast mammographic density using three-dimensional T1-weighted magnetic resonance imaging. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 2268-74	4	68
291	Inherited predisposition to colorectal adenomas caused by multiple rare alleles of MUTYH but not OGG1, NUDT1, NTH1 or NEIL 1, 2 or 3. *Gut*, **2008**, 57, 1252-5	19.2	46

290	Better life expectancy in women with BRCA2 compared with BRCA1 mutations is attributable to lower frequency and later onset of ovarian cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2008**, 17, 1535-42	4	14
289	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
288	Impaired tamoxifen metabolism reduces survival in familial breast cancer patients. *Clinical Cancer Research*, **2008**, 14, 5913-8	12.9	99
287	What are the implications in individuals with unilateral vestibular schwannoma and other neurogenic tumors?. *Journal of Neurosurgery*, **2008**, 108, 92-6	3.2	29
286	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
285	BRCA1/2 mutation analysis in male breast cancer families from North West England. *Familial Cancer* , **2008**, 7, 113-7	3	27
284	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
283	Cost analysis of biomarker testing for mismatch repair deficiency in node-positive colorectal cancer. *British Journal of Surgery*, **2008**, 95, 868-75	5.3	17
282	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
281	BRCA2: a cause of Li-Fraumeni-like syndrome. *Journal of Medical Genetics*, **2008**, 45, 62-3	5.8	15
280	Is CHEK2 a cause of the Li-Fraumeni syndrome?. *Journal of Medical Genetics*, **2008**, 45, 63-4	5.8	22
279	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
278	Characterization of troponin responses in isoproterenol-induced cardiac injury in the Hanover Wistar rat. *Toxicologic Pathology*, **2007**, 35, 606-17	2.1	61
277	Desmoid tumours in patients with familial adenomatous polyposis and desmoid region adenomatous polyposis coli mutations. *British Journal of Surgery*, **2007**, 94, 1009-13	5.3	41
276	Surveillance for familial breast cancer: Differences in outcome according to BRCA mutation status. *International Journal of Cancer*, **2007**, 121, 1017-20	7.5	76
275	PALB2, which encodes a BRCA2-interacting protein, is a breast cancer susceptibility gene. *Nature Genetics*, **2007**, 39, 165-7	36.3	719
274	Predictive genetic testing for BRCA1/2 in a UK clinical cohort: three-year follow-up. *British Journal of Cancer*, **2007**, 96, 718-24	8.7	66
273	Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **2007**, 447, 1087-93	34.4	1957

272	Should NF2 mutation screening be undertaken in patients with an apparently isolated vestibular schwannoma?. *Clinical Genetics*, **2007**, 71, 354-8	4	40
271	The impact of new screening protocol on individuals at increased risk of colorectal cancer. *Colorectal Disease*, **2007**, 9, 635-40	2.1	4
270	Bilateral prophylactic oophorectomy and bilateral prophylactic mastectomy in a prospective cohort of unaffected BRCA1 and BRCA2 mutation carriers. *Clinical Breast Cancer*, **2007**, 7, 875-82	3	69
269	Strategies for identifying hereditary nonpolyposis colon cancer. *Seminars in Oncology*, **2007**, 34, 411-7	5.5	19
268	Familial colorectal cancer referral to regional genetics department--a single centre experience. *Familial Cancer*, **2007**, 6, 81-7	3	5
267	Adult weight gain and central obesity in women with and without a family history of breast cancer: a case control study. *Familial Cancer*, **2007**, 6, 287-94	3	9
266	Delivering cancer genetics services--new ways of working. *Familial Cancer*, **2007**, 6, 163-7	3	14
265	The cost-utility of magnetic resonance imaging for breast cancer in BRCA1 mutation carriers aged 30-49. *European Journal of Health Economics*, **2007**, 8, 137-44	3.6	25
264	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
263	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
262	Cryopreservation of epididymal alpaca (Vicugna pacos) sperm: a comparison of citrate-, Tris- and lactose-based diluents and pellets and straws. *Reproduction, Fertility and Development*, **2007**, 19, 792-6	1.8	31
261	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
260	Breast cancer risk-assessment models. *Breast Cancer Research*, **2007**, 9, 213	8.3	90
259	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes. *Science*, **2007**, 316, 1336-41	33.3	1823
258	Effect of seminal plasma fractions from entire and vasectomized rams on the motility characteristics, membrane status, and in vitro fertility of ram spermatozoa. *Journal of Andrology*, **2007**, 28, 109-22		18
257	Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *Journal of Medical Genetics*, **2007**, 44, 81-8	5.8	600
256	Radiation-induced brain tumours in nevoid basal cell carcinoma syndrome: implications for treatment and surveillance. *Child's Nervous System*, **2007**, 23, 133-6	1.7	20
255	Estrogen deprivation for breast cancer prevention. *Recent Results in Cancer Research*, **2007**, 174, 151-67	1.5	6

254	Extending the Limits of Supertree Methods. *Annals of Combinatorics*, **2006**, 10, 31-51	0.7	7
253	A genome wide linkage search for breast cancer susceptibility genes. *Genes Chromosomes and Cancer*, **2006**, 45, 646-55	5	100
252	The heterogeneous nature of germline mutations in NF1 patients with malignant peripheral serve sheath tumours (MPNSTs). *Human Mutation*, **2006**, 27, 716	4.7	41
251	Evaluation of RAD50 in familial breast cancer predisposition. *International Journal of Cancer*, **2006**, 118, 2911-6	7.5	47
250	Late toxicity is not increased in BRCA1/BRCA2 mutation carriers undergoing breast radiotherapy in the United Kingdom. *Clinical Cancer Research*, **2006**, 12, 7025-32	12.9	59
249	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
248	Optimal selection of individuals for BRCA mutation testing. *Journal of Clinical Oncology*, **2006**, 24, 3311; author reply 3311-2	2.2	1
247	Increasing the specificity of diagnostic criteria for schwannomatosis. *Neurology*, **2006**, 66, 730-2	6.5	121
246	Acute chemotherapy-related toxicity is not increased in BRCA1 and BRCA2 mutation carriers treated for breast cancer in the United Kingdom. *Clinical Cancer Research*, **2006**, 12, 7033-8	12.9	32
245	Is it time to abandon microsatellite instability as a pre-screen for selecting families for mutation testing for mismatch repair genes?. *Journal of Clinical Oncology*, **2006**, 24, 1960-2; author reply 1962-3	2.2	9
244	BRCA1 and BRCA2 cancer risks. *Journal of Clinical Oncology*, **2006**, 24, 3312-3; author reply 3313-4	2.2	23
243	Mammographic density and breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Research*, **2006**, 66, 1866-72	10.1	111
242	Evidence for a colorectal cancer susceptibility locus on chromosome 3q21-q24 from a high-density SNP genome-wide linkage scan. *Human Molecular Genetics*, **2006**, 15, 2903-10	5.6	50
241	Familial ovarian cancer screening. *Journal of Clinical Oncology*, **2006**, 24, e11	2.2	
240	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
239	Evidence of linkage to chromosome 9q22.33 in colorectal cancer kindreds from the United Kingdom. *Cancer Research*, **2006**, 66, 5003-6	10.1	50
238	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
237	Accurate prediction of BRCA1 and BRCA2 heterozygous genotype using expression profiling after induced DNA damage. *Clinical Cancer Research*, **2006**, 12, 3896-901	12.9	32

236	Effects of oestrogens and anti-oestrogens on normal breast tissue from women bearing BRCA1 and BRCA2 mutations. *British Journal of Cancer*, **2006**, 94, 1021-8	8.7	20
235	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
234	A case of multiple cutaneous schwannomas; schwannomatosis or neurofibromatosis type 2?. *Journal of Neurology, Neurosurgery and Psychiatry*, **2006**, 77, 269-71	5.5	21
233	Parity and breast cancer risk among BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2006**, 8, R72	8.3	54
232	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
231	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
230	Screening younger women with a family history of breast cancer--does early detection improve outcome?. *European Journal of Cancer*, **2006**, 42, 1385-90	7.5	38
229	Psychosocial effects of neurofibromatosis type 2 (Part 1): General effects. *Audiological Medicine*, **2006**, 4, 202-210		7
228	Predicting compliance in a breast cancer prevention trial. *Breast Journal*, **2006**, 12, 446-50	1.2	16
227	Inherited association of breast and colorectal cancer: limited role of CHEK2 compared with high-penetrance genes. *Clinical Genetics*, **2006**, 70, 388-95	4	16
226	ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. *Nature Genetics*, **2006**, 38, 873-5	36.3	553
225	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
224	News on the genetics, epidemiology, medical care and translational research of Schwannomas. *Journal of Neurology*, **2006**, 253, 1533-41	5.5	41
223	The development and evaluation of alternative communication strategies to facilitate interactions with Somali refugees in primary care: a preliminary study. *Journal of Innovation in Health Informatics* , **2006**, 14, 183-9		2
222	Family history of breast cancer: referral guidelines changed after acceptance of 10 minute consultation. *BMJ, The*, **2005**, 330, 730; author reply 730	5.9	1
221	Sensitive detection of deletions of one or more exons in the neurofibromatosis type 2 (NF2) gene by multiplexed gene dosage polymerase chain reaction. *Journal of Molecular Diagnostics*, **2005**, 7, 97-104	5.1	10
220	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
219	Surgical decisions made by 158 women with hereditary breast cancer aged . *European Journal of Surgical Oncology*, **2005**, 31, 1112-8	3.6	37

218	Mechanisms of Disease: prediction and prevention of breast cancer--cellular and molecular interactions. *Nature Clinical Practice Oncology*, **2005**, 2, 635-46		24
217	Size and growth rate of sporadic vestibular schwannoma: predictive value of information available at presentation. *Otology and Neurotology*, **2005**, 26, 86-92	2.6	52
216	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
215	Identification of genetic aberrations on chromosome 22 outside the NF2 locus in schwannomatosis and neurofibromatosis type 2. *Human Mutation*, **2005**, 26, 540-9	4.7	25
214	High-resolution array-CGH profiling of germline and tumor-specific copy number alterations on chromosome 22 in patients affected with schwannomas. *Human Genetics*, **2005**, 118, 35-44	6.3	18
213	Neurofibromatosis 2 (NF2) and malignant mesothelioma in a man with a constitutional NF2 missense mutation. *Familial Cancer*, **2005**, 4, 321-2	3	17
212	Multiple meningiomas: differential involvement of the NF2 gene in children and adults. *Journal of Medical Genetics*, **2005**, 42, 45-8	5.8	49
211	Diagnostic criteria for schwannomatosis. *Neurology*, **2005**, 64, 1838-45	6.5	309
210	Non-random transmission of mutant alleles to female offspring in BRCA carriers. *Journal of Medical Genetics*, **2005**, 42, e6	5.8	3
209	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
208	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
207	Spinal tumors in neurofibromatosis type 2. Is emerging knowledge of genotype predictive of natural history?. *Journal of Neurosurgery: Spine*, **2005**, 2, 574-9	2.8	54
206	Methodological issues in longitudinal studies: vestibular schwannoma growth rates in neurofibromatosis 2. *Journal of Medical Genetics*, **2005**, 42, 903-6	5.8	24
205	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
204	Evidence for an association between compound heterozygosity for germ line mutations in the hemochromatosis (HFE) gene and increased risk of colorectal cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2005**, 14, 1460-3	4	20
203	The location of constitutional neurofibromatosis 2 (NF2) splice site mutations is associated with the severity of NF2. *Journal of Medical Genetics*, **2005**, 42, 540-6	5.8	72
202	Update on the Manchester Scoring System for BRCA1 and BRCA2 testing. *Journal of Medical Genetics*, **2005**, 42, e39	5.8	60
201	Age related shift in the mutation spectra of germline and somatic NF2 mutations: hypothetical role of DNA repair mechanisms. *Journal of Medical Genetics*, **2005**, 42, 630-2	5.8	24

200	Management of the patient and family with neurofibromatosis 2: a consensus conference statement. *British Journal of Neurosurgery*, **2005**, 19, 5-12	1	187
199	Neurofibromatosis type 2 **2004**, 50-59		0
198	Mutation scanning of the NF2 gene: an improved service based on meta-PCR/sequencing, dosage analysis, and loss of heterozygosity analysis. *Genetic Testing and Molecular Biomarkers*, **2004**, 8, 368-80		31
197	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
196	Constitutional rearrangements of chromosome 22 as a cause of neurofibromatosis 2. *Journal of Medical Genetics*, **2004**, 41, 529-34	5.8	39
195	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
194	Non-uptake of predictive genetic testing for BRCA1/2 among relatives of known carriers: attributes, cancer worry, and barriers to testing in a multicenter clinical cohort. *Genetic Testing and Molecular Biomarkers*, **2004**, 8, 23-9		52
193	Haplotype and cancer risk analysis of two common mutations, BRCA1 4184del4 and BRCA2 2157delG, in high risk northwest England breast/ovarian families. *Journal of Medical Genetics*, **2004**, 41, e21	5.8	15
192	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
191	BRCA1/2 predictive testing: a study of uptake in two centres. *European Journal of Human Genetics*, **2004**, 12, 654-62	5.3	44
190	Prevention and genetic testing for breast cancer: variations in medical decisions. *Social Science and Medicine*, **2004**, 58, 1085-96	5.1	39
189	Molecular stool screening for colorectal cancer. *British Journal of Surgery*, **2004**, 91, 790-800	5.3	26
188	Eight novel MSH6 germline mutations in patients with familial and nonfamilial colorectal cancer selected by loss of protein expression in tumor tissue. *Human Mutation*, **2004**, 23, 285	4.7	26
187	Are BRCA1- and BRCA2-related breast cancers associated with increased mortality?. *Breast Cancer Research*, **2004**, 6, E7	8.3	17
186	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
185	Somatic mosaicism in neurofibromatosis 2: prevalence and risk of disease transmission to offspring. *Journal of Medical Genetics*, **2003**, 40, 459-63	5.8	103
184	Neurofibromatosis 2. *Current Opinion in Neurology*, **2003**, 16, 27-33	7.1	93
183	Patterns of associations of clinical features in neurofibromatosis 1 (NF1). *Human Genetics*, **2003**, 112, 289-97	6.3	43

182	New approaches to the endocrine prevention and treatment of breast cancer. *Cancer Chemotherapy and Pharmacology*, **2003**, 52 Suppl 1, S39-44	3.5	26
181	Exploring the "two-hit hypothesis" in NF2: tests of two-hit and three-hit models of vestibular schwannoma development. *Genetic Epidemiology*, **2003**, 24, 265-72	2.6	21
180	Dento-osseous changes as diagnostic markers in familial adenomatous polyposis families. *Oral Diseases*, **2003**, 9, 29-33	3.5	9
179	Genetic analysis of mitochondrial complex II subunits SDHD, SDHB and SDHC in paraganglioma and phaeochromocytoma susceptibility. *Clinical Endocrinology*, **2003**, 59, 728-33	3.4	81
178	Is clinical growth index a reliable predictor of tumour growth in vestibular schwannomas?. *Clinical Otolaryngology*, **2003**, 28, 85-90		26
177	Bayesian evaluation of breast cancer screening using data from two studies. *Statistics in Medicine*, **2003**, 22, 1661-74	2.3	7
176	Prediction of pathogenic mutations in patients with early-onset breast cancer by family history. *Lancet, The*, **2003**, 361, 1101-2	40	164
175	Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. *Lancet, The*, **2003**, 362, 39-41	40	358
174	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
173	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
172	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
171	Abnormalities of the vitreoretinal interface caused by dysregulated Hedgehog signaling during retinal development. *Human Molecular Genetics*, **2003**, 12, 3269-76	5.6	38
170	Genotype-phenotype correlations for cataracts in neurofibromatosis 2. *Journal of Medical Genetics*, **2003**, 40, 758-60	5.8	12
169	Identification of recurrent regions of chromosome loss and gain in vestibular schwannomas using comparative genomic hybridisation. *Journal of Medical Genetics*, **2003**, 40, 802-6	5.8	56
168	Sensitivity of BRCA1/2 mutation testing in 466 breast/ovarian cancer families. *Journal of Medical Genetics*, **2003**, 40, e107	5.8	9
167	Do women understand the odds? Risk perceptions and recall of risk information in women with a family history of breast cancer. *Public Health Genomics*, **2003**, 6, 214-23	1.9	25
166	Evaluation of Fanconi Anemia genes in familial breast cancer predisposition. *Cancer Research*, **2003**, 63, 8596-9	10.1	41
165	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

164	Intrafamilial correlation of clinical manifestations in neurofibromatosis 2 (NF2). *Genetic Epidemiology*, **2002**, 23, 245-59	2.6	20
163	Contribution of cyclin d1 (CCND1) and E-cadherin (CDH1) polymorphisms to familial and sporadic colorectal cancer. *Oncogene*, **2002**, 21, 1928-33	9.2	80
162	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
161	A follow-up study of breast and other cancers in families of an unselected series of breast cancer patients. *British Journal of Cancer*, **2002**, 86, 718-22	8.7	11
160	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
159	Re: risk-reduction mastectomy: clinical issues and research needs. *Journal of the National Cancer Institute*, **2002**, 94, 307-8	9.7	3
158	Prophylactic oophorectomy in carriers of BRCA1 or BRCA2 mutations. *New England Journal of Medicine*, **2002**, 346, 1616-22	59.2	1363
157	Neurofibromatosis type 1 and sporadic optic gliomas. *Archives of Disease in Childhood*, **2002**, 87, 65-70	2.2	113
156	Risk assessment and management of high risk familial breast cancer. *Journal of Medical Genetics*, **2002**, 39, 865-71	5.8	43
155	Evaluation of linkage of breast cancer to the putative BRCA3 locus on chromosome 13q21 in 128 multiple case families from the Breast Cancer Linkage Consortium. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 827-31	11.5	64
154	Germline mutation of ARF in a melanoma kindred. *Human Molecular Genetics*, **2002**, 11, 1273-9	5.6	107
153	Evaluation of clinical diagnostic criteria for neurofibromatosis 2. *Neurology*, **2002**, 59, 1759-65	6.5	185
152	Molecular genetic analysis of the NF2 gene in young patients with unilateral vestibular schwannomas. *Journal of Medical Genetics*, **2002**, 39, 315-22	5.8	52
151	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
150	Predictors of the risk of mortality in neurofibromatosis 2. *American Journal of Human Genetics*, **2002**, 71, 715-23	11	178
149	Screening of patients at high risk of colorectal cancer. *Colorectal Disease*, **2001**, 3, 308-11	2.1	4
148	Women's attitudes toward preventive strategies for hereditary breast or ovarian carcinoma differ from one country to another: differences among English, French, and Canadian women. *Cancer*, **2001**, 92, 959-68	6.4	83
147	High detection rate for BRCA2 mutations in male breast cancer families from North West England. *Familial Cancer*, **2001**, 1, 131-3	3	10

146	Familial breast cancer: an investigation into the outcome of treatment for early stage disease. *Familial Cancer*, **2001**, 1, 65-72	3	39
145	Relative frequency and morphology of cancers in carriers of germline TP53 mutations. *Oncogene*, **2001**, 20, 4621-8	9.2	361
144	Molecular genetics and endometrial cancer. *Best Practice and Research in Clinical Obstetrics and Gynaecology*, **2001**, 15, 355-63	4.6	4
143	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
142	Germline mutation analysis of the transforming growth factor beta receptor type II (TGFBR2) and E-cadherin (CDH1) genes in early onset and familial colorectal cancer. *Journal of Medical Genetics*, **2001**, 38, E7	5.8	3
141	Re: Characterization of hereditary nonpolyposis colorectal cancer families from a population-based series of cases. *Journal of the National Cancer Institute*, **2001**, 93, 716-7	9.7	6
140	There may never be a final cure for breast cancer. *European Journal of Surgical Oncology*, **2001**, 27, 338-9	3.6	2
139	Germline SDHD mutation in familial phaeochromocytoma. *Lancet, The*, **2001**, 357, 1181-2	40	211
138	Uptake of screening and prevention in women at very high risk of breast cancer. *Lancet, The*, **2001**, 358, 889-90	40	48
137	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
136	Advances in neurofibromatosis 2 (NF2): a workshop report. *Journal of Neurogenetics*, **2000**, 14, 63-106	1.6	26
135	Lack of sex-ratio distortion in neurofibromatosis 2. *American Journal of Medical Genetics Part A*, **2000**, 95, 292-292		1
134	Associations of clinical features in neurofibromatosis 1 (NF1). *Genetic Epidemiology*, **2000**, 19, 429-39	2.6	66
133	Clinical follow-up after bilateral risk reducing ('prophylactic') mastectomy: mental health and body image outcomes. *Psycho-Oncology*, **2000**, 9, 462-72	3.9	103
132	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
131	Identification of the familial cylindromatosis tumour-suppressor gene. *Nature Genetics*, **2000**, 25, 160-5	36.3	560
130	No useful role for fine needle aspiration as a marker for familial breast cancer. *Breast*, **2000**, 9, 218-9	3.6	
129	Rationale for a national multi-centre study of magnetic resonance imaging screening in women at genetic risk of breast cancer. *Breast*, **2000**, 9, 72-7	3.6	16

128	Protocol for a national multi-centre study of magnetic resonance imaging screening in women at genetic risk of breast cancer. *Breast*, **2000**, 9, 78-82	3.6	21
127	Management of the contralateral breast in patients with hereditary breast cancer. *Breast*, **2000**, 9, 301-5	3.6	2
126	Neurofibromatosis 2, radiosurgery and malignant nervous system tumours. *British Journal of Cancer* , **2000**, 82, 998	8.7	101
125	Linkage and LOH studies in 19 cylindromatosis families show no evidence of genetic heterogeneity and refine the CYLD locus on chromosome 16q12-q13. *Human Genetics*, **2000**, 106, 58-65	6.3	46
124	A survey of the current clinical facilities for the management of familial cancer in Europe. European Union BIOMED II Demonstration Project: Familial Breast Cancer: audit of a new development in medical practice in European centres. *Journal of Medical Genetics*, **2000**, 37, 605-7	5.8	13
123	Guidelines for a genetic risk based approach to advising women with a family history of breast cancer. UK Cancer Family Study Group (UKCFSG). *Journal of Medical Genetics*, **2000**, 37, 203-9	5.8	80
122	Can hair be used to screen for breast cancer?. *Journal of Medical Genetics*, **2000**, 37, 297-8	5.8	13
121	Analysis of genetic and phenotypic heterogeneity in juvenile polyposis. *Gut*, **2000**, 46, 656-60	19.2	95
120	Clinical and molecular correlates of somatic mosaicism in neurofibromatosis 2. *Journal of Medical Genetics*, **2000**, 37, 542-3	5.8	3
119	Recurrent germline mutation in MSH2 arises frequently de novo. *Journal of Medical Genetics*, **2000**, 37, 646-52	5.8	63
118	2157delG: a frequent mutation in BRCA2 missed by PTT. *Journal of Medical Genetics*, **2000**, 37, E42	5.8	6
117	Use of MRI and audiological tests in presymptomatic diagnosis of type 2 neurofibromatosis (NF2). *Journal of Medical Genetics*, **2000**, 37, 944-7	5.8	26
116	Variation in prophylactic surgery decisions. *Lancet, The*, **2000**, 356, 1687	40	9
115	A protocol for preventative mastectomy in women with an increased lifetime risk of breast cancer. *European Journal of Surgical Oncology*, **2000**, 26, 711-3	3.6	27
114	Hereditary cancer. *Lancet Oncology, The*, **2000**, 1, 12-3	21.7	
113	Neurofibromatosis type 2. *Journal of Medical Genetics*, **2000**, 37, 897-904	5.8	128
112	Genetic and cytogenetic studies in inherited cancer: Li-Fraumeni syndrome **2000**, 245-255		
111	Guidelines for follow-up of women at high risk for inherited breast cancer: consensus statement from the Biomed 2 Demonstration Programme on Inherited Breast Cancer. *Disease Markers*, **1999**, 15, 207-11	3.2	54

110	Cancer genetics services in Europe. *Disease Markers*, **1999**, 15, 3-13	3.2	12
109	Insurance implications for individuals with a high risk of breast and ovarian cancer in Europe. *Disease Markers*, **1999**, 15, 159-65	3.2	10
108	Genetic testing for breast cancer predisposition in 1999: which molecular strategy and which family criteria?. *Disease Markers*, **1999**, 15, 67-8	3.2	4
107	Ethical, social and economic issues in familial breast cancer: a compilation of views from the E.C. Biomed II Demonstration Project. *Disease Markers*, **1999**, 15, 125-31	3.2	23
106	Utilisation of prophylactic mastectomy in 10 European centres. *Disease Markers*, **1999**, 15, 148-51	3.2	27
105	Efficacy of early diagnosis and treatment in women with a family history of breast cancer. European Familial Breast Cancer Collaborative Group. *Disease Markers*, **1999**, 15, 179-86	3.2	34
104	Management of hereditary breast cancer. European Familial Breast Cancer Collaborative Group. *Disease Markers*, **1999**, 15, 187-9	3.2	6
103	Risk estimation as a decision-making tool for genetic analysis of the breast cancer susceptibility genes. EC Demonstration Project on Familial Breast Cancer. *Disease Markers*, **1999**, 15, 53-65	3.2	7
102	Neurofibromatosis Type 2: Genetic and Clinical Features. *Ear, Nose and Throat Journal*, **1999**, 78, 97-100	1	20
101	Germline E-cadherin gene (CDH1) mutations predispose to familial gastric cancer and colorectal cancer. *Human Molecular Genetics*, **1999**, 8, 607-10	5.6	271
100	Probability of bilateral disease in people presenting with a unilateral vestibular schwannoma. *Journal of Neurology, Neurosurgery and Psychiatry*, **1999**, 66, 764-7	5.5	46
99	Paediatric presentation of type 2 neurofibromatosis. *Archives of Disease in Childhood*, **1999**, 81, 496-9	2.2	156
98	Differences among Helicobacter pylori strains isolated from three different populations and demonstrated by restriction enzyme analysis of an internal fragment of the conserved gene hpaA. *Helicobacter*, **1999**, 4, 82-8	4.9	8
97	The pathology of familial breast cancer: Clinical and genetic counselling implications of breast cancer pathology. *Breast Cancer Research*, **1999**, 1, 48-51	8.3	8
96	The Fanconi anemia group E gene, FANCE, maps to chromosome 6p. *American Journal of Human Genetics*, **1999**, 64, 1400-5	11	43
95	Are there low-penetrance TP53 Alleles? evidence from childhood adrenocortical tumors. *American Journal of Human Genetics*, **1999**, 65, 995-1006	11	211
94	The genetic epidemiology of early-onset epithelial ovarian cancer: a population-based study. *American Journal of Human Genetics*, **1999**, 65, 1725-32	11	60
93	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

92	A common MSH2 mutation in English and North American HNPCC families: origin, phenotypic expression, and sex specific differences in colorectal cancer. *Journal of Medical Genetics*, **1999**, 36, 97-102	5.8	59
91	A clinical study of type 1 neurofibromatosis in north west England. *Journal of Medical Genetics*, **1999**, 36, 197-203	5.8	121
90	The accuracy of diagnoses as reported in families with cancer: a retrospective study. *Journal of Medical Genetics*, **1999**, 36, 309-12	5.8	74
89	Mononucleotide microsatellite instability and germline MSH6 mutation analysis in early onset colorectal cancer. *Journal of Medical Genetics*, **1999**, 36, 678-82	5.8	55
88	Genetic and functional studies of a germline TP53 splicing mutation in a Li-Fraumeni-like family. *Oncogene*, **1998**, 16, 3291-8	9.2	41
87	Cancer phenotype correlates with constitutional TP53 genotype in families with the Li-Fraumeni syndrome. *Oncogene*, **1998**, 17, 1061-8	9.2	160
86	A novel TP53 splicing mutation in a Li-Fraumeni syndrome family: a patient with Wilms' tumour is not a mutation carrier. *British Journal of Cancer*, **1998**, 78, 1081-3	8.7	13
85	Psychological support needs for women at high genetic risk of breast cancer: some preliminary indicators. *Psycho-Oncology*, **1998**, 7, 402-12	3.9	89
84	Effect of early American results on patients in a tamoxifen prevention trial (IBIS). International Breast Cancer Intervention Study. *Lancet, The*, **1998**, 352, 1222	40	8
83	Molecular genetic tests in surgical management of familial adenomatous polyposis. *Lancet, The*, **1998**, 351, 1131-1132	40	5
82	Screening by mammography, women with a family history of breast cancer. *European Journal of Cancer*, **1998**, 34, 937-40	7.5	74
81	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
80	False family history of breast cancer in the family cancer clinic. *European Journal of Surgical Oncology*, **1998**, 24, 275-9	3.6	18
79	Somatic mosaicism: a common cause of classic disease in tumor-prone syndromes? Lessons from type 2 neurofibromatosis. *American Journal of Human Genetics*, **1998**, 63, 727-36	11	35
78	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
77	An evaluation of common breast cancer gene mutations in a population of Ashkenazi Jews. *Journal of Medical Genetics*, **1998**, 35, 10-2	5.8	6
76	Differential diagnosis of type 2 neurofibromatosis: molecular discrimination of NF2 and sporadic vestibular schwannomas. *Journal of Medical Genetics*, **1998**, 35, 973-7	5.8	16
75	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

74	Men in breast cancer families: a preliminary qualitative study of awareness and experience. *Journal of Medical Genetics*, **1998**, 35, 739-44	5.8	53
73	A system for enabling blind people to identify landmarks: the sound buoy. *IEEE Transactions on Rehabilitation Engineering: A Publication of the IEEE Engineering in Medicine and Biology Society*, **1997**, 5, 276-8		18
72	Physical localisation of the breakpoints of a constitutional translocation t(5;6)(q21;q21) in a child with bilateral Wilms' tumour. *Journal of Medical Genetics*, **1997**, 34, 343-5	5.8	15
71	Uptake of genetic testing for cancer predisposition. *Journal of Medical Genetics*, **1997**, 34, 746-8	5.8	50
70	Spinal and cutaneous schwannomatosis is a variant form of type 2 neurofibromatosis: a clinical and molecular study. *Journal of Neurology, Neurosurgery and Psychiatry*, **1997**, 62, 361-6	5.5	53
69	Misleading linkage results in an NF2 presymptomatic test owing to mosaicism. *Journal of Medical Genetics*, **1997**, 34, 934-6	5.8	19
68	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
67	Molecular genetic tests in surgical management of familial adenomatous polyposis. *Lancet, The*, **1997**, 350, 1777	40	12
66	A detailed study of loss of heterozygosity on chromosome 17 in tumours from Li-Fraumeni patients carrying a mutation to the TP53 gene. *Oncogene*, **1997**, 14, 865-71	9.2	88
65	Li-Fraumeni syndrome--a molecular and clinical review. *British Journal of Cancer*, **1997**, 76, 1-14	8.7	274
64	Risk of subsequent primary cancers in patients with carcinoma of the Ampulla of Vater. *British Journal of Cancer*, **1997**, 76, 1232-3	8.7	2
63	APC mutations in familial adenomatous polyposis families in the Northwest of England. *Human Mutation*, **1997**, 10, 376-80	4.7	37
62	Germ-line mutations of TP53 in Li-Fraumeni families: an extended study of 39 families. *Cancer Research*, **1997**, 57, 3245-52	10.1	169
61	Microsatellite instability in early onset and familial colorectal cancer. *Journal of Medical Genetics*, **1996**, 33, 981-5	5.8	29
60	Neurofibromatosis/Noonan phenotype: a variable feature of type 1 neurofibromatosis. *Clinical Genetics*, **1996**, 49, 59-64	4	57
59	Familial infiltrative fibromatosis (desmoid tumours) (MIM135290) caused by a recurrent 3' APC gene mutation. *Human Molecular Genetics*, **1996**, 5, 1921-4	5.6	108
58	MSH2 sequence variations and inherited colorectal cancer susceptibility. *European Journal of Cancer*, **1996**, 32A, 178	7.5	10
57	Cancer genetics clinics. *European Journal of Cancer*, **1996**, 32A, 391-2	7.5	20

56	Fictitious breast cancer family history. *Lancet, The*, **1996**, 348, 1034	40	15
55	A clinical, genetic and audiological study of patients and families with unilateral vestibular schwannomas. I. Clinical features of neurofibromatosis in patients with unilateral vestibular schwannomas. *Journal of Laryngology and Otology*, **1996**, 110, 634-640	1.8	14
54	A novel deletion within exon 6 of TP53 in a family with Li-Fraumeni-like syndrome, and LOH in a benign lesion from a mutation carrier. *Cancer Genetics and Cytogenetics*, **1996**, 90, 14-6		11
53	Genetic markers for breast cancer. *Breast*, **1996**, 5, 374-376	3.6	1
52	Florid oral manifestations in an atypical familial adenomatous polyposis family with late presentation of colorectal polyps. *Journal of Oral Pathology and Medicine*, **1996**, 25, 459-62	3.3	2
51	Mutation screening of MSH2 and MLH1 mRNA in hereditary non-polyposis colon cancer syndrome. *Journal of Medical Genetics*, **1996**, 33, 726-30	5.8	40
50	A genetic register for von Hippel-Lindau disease. *Journal of Medical Genetics*, **1996**, 33, 120-7	5.8	169
49	The ethics of testing for cancer-predisposition genes **1996**, 383-393		1
48	A clinical, genetic and audiological study of patients and families with unilateral vestibular schwannomas. I. Clinical features of neurofibromatosis in patients with unilateral vestibular schwannomas. *Journal of Laryngology and Otology*, **1996**, 110, 634-40	1.8	6
47	Molecular genetic analysis of exons 1 to 6 of the APC gene in non-polyposis familial colorectal cancer. *Clinical Genetics*, **1995**, 48, 299-303	4	6
46	Eleven novel mutations in the NF2 tumour suppressor gene. *Human Genetics*, **1995**, 95, 572-4	6.3	15
45	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
44	Should I Take HRT, Doctor? Hormone Replacement Therapy in Women at Increased Risk of Breast Cancer and in Survivors of the Disease. *The Journal of the British Menopause Society*, **1995**, 1, 9-17		2
43	Suggested screening guidelines for familial colorectal cancer. *Journal of Medical Screening*, **1995**, 2, 45-51	1.4	20
42	An extended Li-Fraumeni kindred with gastric carcinoma and a codon 175 mutation in TP53. *Journal of Medical Genetics*, **1995**, 32, 942-5	5.8	55
41	WAGR syndrome and multiple exostoses in a patient with del(11)(p11.2p14.2). *Journal of Medical Genetics*, **1995**, 32, 823-4	5.8	24
40	Diagnostic issues in a family with late onset type 2 neurofibromatosis. *Journal of Medical Genetics*, **1995**, 32, 470-4	5.8	19
39	Genetic linkage analysis in hereditary non-polyposis colon cancer syndrome. *Journal of Medical Genetics*, **1995**, 32, 352-7	5.8	22

38	Genetic evidence for host specificity in the adhesin-encoding genes hxaA of Helicobacter acinonyx, hnaA of H. nemestrinae and hpaA of H. pylori. *Gene*, **1995**, 163, 97-102	3.8	9
37	Variation of expression of the gene for type 2 neurofibromatosis: absence of a gender effect on vestibular schwannomas, but confirmation of a preponderance of meningiomas in females. *Journal of Laryngology and Otology*, **1995**, 109, 830-5	1.8	17
36	Non-expression of von Hippel-Lindau phenotype in an obligate gene carrier. *Clinical Genetics*, **1994**, 45, 104-6	4	7
35	Germline mutations in the neurofibromatosis type 2 tumour suppressor gene. *Human Molecular Genetics*, **1994**, 3, 813-6	5.6	100
34	Detailed mapping of germline deletions of the von Hippel-Lindau disease tumour suppressor gene. *Human Molecular Genetics*, **1994**, 3, 595-8	5.6	71
33	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
32	Apo E genotypes in multiple sclerosis, Parkinson's disease, schwannomas and late-onset Alzheimer's disease. *Molecular and Cellular Probes*, **1994**, 8, 519-25	3.3	54
31	Familial breast cancer. *BMJ: British Medical Journal*, **1994**, 308, 183-7		61
30	A mutation in the neurofibromatosis type 2 tumor-suppressor gene, giving rise to widely different clinical phenotypes in two unrelated individuals. *American Journal of Human Genetics*, **1994**, 55, 69-73	11	51
29	Non-penetrance and late appearance of polyps in families with familial adenomatous polyposis. *Gut* , **1993**, 34, 1389-93	19.2	25
28	A disease-associated germline deletion maps the type 2 neurofibromatosis (NF2) gene between the Ewing sarcoma region and the leukaemia inhibitory factor locus. *Human Molecular Genetics*, **1993**, 2, 701-4	5.6	52
27	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
26	Perception of risk in women with a family history of breast cancer. *British Journal of Cancer*, **1993**, 67, 612-4	8.7	147
25	A clinical, genetic and audiological study of patients and families with bilateral acoustic neurofibromatosis. *Journal of Laryngology and Otology*, **1993**, 107, 6-11	1.8	7
24	Type 2 neurofibromatosis: the need for supraregional care?. *Journal of Laryngology and Otology*, **1993**, 107, 401-6	1.8	44
23	Assessment of relative risk of second primary tumors after ovarian cancer and of the usefulness of double primary cases as a source of material for genetic studies with a cancer registry. *Cancer*, **1993** , 72, 819-27	6.4	24
22	Congenital anomalies and genetic syndromes in 173 cases of medulloblastoma. *Medical and Pediatric Oncology*, **1993**, 21, 433-4		33
21	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

20	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
19	Ovarian cancer family and prophylactic choices. *Journal of Medical Genetics*, **1992**, 29, 416-8	5.8	15
18	Location of gene for Gorlin syndrome. *Lancet, The*, **1992**, 339, 581-2	40	336
17	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
16	A clinical study of type 2 neurofibromatosis. *The Quarterly Journal of Medicine*, **1992**, 84, 603-18		323
15	Dominantly inherited microcephaly, hypotelorism and normal intelligence. *Clinical Genetics*, **1991**, 39, 178-80	4	12
14	Cutaneous lymphangioma and amegakaryocytic thrombocytopenia in Noonan syndrome. *Clinical Genetics*, **1991**, 39, 228-32	4	18
13	The incidence of Gorlin syndrome in 173 consecutive cases of medulloblastoma. *British Journal of Cancer*, **1991**, 64, 959-61	8.7	242
12	Fatal congenital cytomegalovirus infection acquired by an intra-uterine transfusion. *European Journal of Pediatrics*, **1991**, 150, 780-1	4.1	18
11	Space infections of the head and neck--the "new" clinical picture. *Journal of the Royal Army Medical Corps*, **1991**, 137, 35-7	0.8	
10	Family implications of neonatal Gorlin's syndrome. *Archives of Disease in Childhood*, **1991**, 66, 1162-3	2.2	15
9	Brain tumours and the occurrence of severe invasive basal cell carcinoma in first degree relatives with Gorlin syndrome. *British Journal of Neurosurgery*, **1991**, 5, 643-6	1	44
8	Heredity and dysmorphic syndromes in congenital limb deficiencies. *Prosthetics and Orthotics International*, **1991**, 15, 70-7	1.5	3
7	Ankyloblepharon filiforme adnatum in trisomy 18 Edwards syndrome. *Journal of Medical Genetics*, **1990**, 27, 720-1	5.8	8
6	Common variants in breast cancer risk loci predispose to distinct tumor subtypes		1
5	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
4	Prospective Evaluation of a Breast Cancer Risk Model Integrating Classical Risk Factors and Polygenic Risk in 15 Cohorts from Six Countries		2
3	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2

2	Characterising the loss-of-function impact of 5′untranslated region variants in whole genome sequence data from 15,708 individuals		5
1	Back to the future? Reflections on three phases of education policy reform in Wales and their implications for teachers. *Journal of Educational Change*,1	1.8	5