# Gareth Evans

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 1,004 papers | 86,902 citations | 356 136 h-index | 642 256 g-index |
| 1056 all docs | 1056 docs citations | 1056 times ranked | 53916 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | 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, 1117-1130. | 6.2 | 3,105 |
| 2 | Genome-wide association study identifies novel breast cancer susceptibility loci. Nature, 2007, 447, 1087-1093. | 27.8 | 2,165 |
| 3 | Replication of Genome-Wide Association Signals in UK Samples Reveals Risk Loci for Type 2 Diabetes. Science, 2007, 316, 1336-1341. | 12.6 | 2,040 |
| 4 | Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers. JAMA - Journal of the American Medical Association, 2017, 317, 2402. | 7.4 | 1,898 |
| 5 | Prophylactic Oophorectomy in Carriers ofBRCA1orBRCA2Mutations. New England Journal of Medicine, 2002, 346, 1616-1622. | 27.0 | 1,565 |
| 6 | Association of Risk-Reducing Surgery in <emph type="ital">BRCA1</emph> or <emph type="ital">BRCA2</emph> Mutation Carriers With Cancer Risk and Mortality. JAMA - Journal of the American Medical Association, 2010, 304, 967. | 7.4 | 1,241 |
| 7 | Bilateral Prophylactic Mastectomy Reduces Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers: The PROSE Study Group. Journal of Clinical Oncology, 2004, 22, 1055-1062. | 1.6 | 1,095 |
| 8 | Low-penetrance susceptibility to breast cancer due to CHEK2*1100delC in noncarriers of BRCA1 or BRCA2 mutations. Nature Genetics, 2002, 31, 55-59. | 21.4 | 1,001 |
| 9 | Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups (update from 2002). Gut, 2010, 59, 666-689. | 12.1 | 1,000 |
| 10 | PALB2, which encodes a BRCA2-interacting protein, is a breast cancer susceptibility gene. Nature Genetics, 2007, 39, 165-167. | 21.4 | 858 |
| 11 | 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, 2081-2087. | 13.7 | 849 |
| 12 | Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. Journal of Medical Genetics, 2006, 44, 81-88. | 3.2 | 778 |
| 13 | Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257. | 27.0 | 764 |
| 14 | Cancer Risks for BRCA1 and BRCA2 Mutation Carriers: Results From Prospective Analysis of EMBRACE. Journal of the National Cancer Institute, 2013, 105, 812-822. | 6.3 | 753 |
| 15 | Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720. | 27.8 | 737 |
| 16 | Birth incidence and prevalence of tumorâ€prone syndromes: Estimates from a UK family genetic register service. American Journal of Medical Genetics, Part A, 2010, 152A, 327-332. | 1.2 | 721 |
| 17 | Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34. | 6.2 | 711 |
| 18 | Genome-wide association study identifies five new breast cancer susceptibility loci. Nature Genetics, 2010, 42, 504-507. | 21.4 | 653 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. Nature Genetics, 2006, 38, 873-875. | 21.4 | 641 |
| 20 | 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, 1748-1757. | 1.6 | 641 |
| 21 | Identification of the familial cylindromatosis tumour-suppressor gene. Nature Genetics, 2000, 25, 160-165. | 21.4 | 640 |
| 22 | Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. Nature Genetics, 2006, 38, 1239-1241. | 21.4 | 636 |
| 23 | Oral Contraceptives and the Risk of Hereditary Ovarian Cancer. New England Journal of Medicine, 1998, 339, 424-428. | 27.0 | 591 |
| 24 | Prediction of BRCA1 Status in Patients with Breast Cancer Using Estrogen Receptor and Basal Phenotype. Clinical Cancer Research, 2005, 11, 5175-5180. | 7.0 | 577 |
| 25 | 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, 714-727. | 3.4 | 573 |
| 26 | Association Between <emph type="ital">BRCA1</emph> and <emph type="ital">BRCA2</emph> Mutations and Survival in Women With Invasive Epithelial Ovarian Cancer. JAMA - Journal of the American Medical Association, 2012, 307, 382. | 7.4 | 546 |
| 27 | Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439. | 27.0 | 532 |
| 28 | 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, 1331-1337. | 1.6 | 522 |
| 29 | A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. Nature Genetics, 2008, 40, 623-630. | 21.4 | 514 |
| 30 | 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, 134-147. | 2.5 | 513 |
| 31 | Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384. | 21.4 | 493 |
| 32 | Complications of the naevoid basal cell carcinoma syndrome: results of a population based study.. Journal of Medical Genetics, 1993, 30, 460-464. | 3.2 | 485 |
| 33 | Germline mutations of the BRCA1 gene in breast and ovarian cancer families provide evidence for a genotypeâ€"phenotype correlation. Nature Genetics, 1995, 11, 428-433. | 21.4 | 484 |
| 34 | The BOADICEA model of genetic susceptibility to breast and ovarian cancers: updates and extensions. British Journal of Cancer, 2008, 98, 1457-1466. | 6.4 | 461 |
| 35 | Germline mutations in RAD51D confer susceptibility to ovarian cancer. Nature Genetics, 2011, 43, 879-882. | 21.4 | 460 |
| 36 | Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. Nature Genetics, 2009, 41, 585-590. | 21.4 | 434 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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, 841-846. | 3.2 | 421 |
| 38 | Autosomal recessive colorectal adenomatous polyposis due to inherited mutations of MYH. Lancet, The, 2003, 362, 39-41. | 13.7 | 421 |
| 39 | Assessing Women at High Risk of Breast Cancer: A Review of Risk Assessment Models. Journal of the National Cancer Institute, 2010, 102, 680-691. | 6.3 | 413 |
| 40 | 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. | 12.1 | 411 |
| 41 | Relative frequency and morphology of cancers in carriers of germline TP53 mutations. Oncogene, 2001, 20, 4621-4628. | 5.9 | 410 |
| 42 | Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut, 2018, 67, 1306-1316. | 12.1 | 410 |
| 43 | Neurofibromatosis type 2 (NF2): A clinical and molecular review. Orphanet Journal of Rare Diseases, 2009, 4, 16. | 2.7 | 404 |
| 44 | 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, 7804-7810. | 1.6 | 396 |
| 45 | Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347. | 7.4 | 390 |
| 46 | Allele losses in the region 17q12â€“21 in familial breast and ovarian cancer involve the wildâ€“type chromosome. Nature Genetics, 1992, 2, 128-131. | 21.4 | 387 |
| 47 | Location of gene for Gorlin syndrome. Lancet, The, 1992, 339, 581-582. | 13.7 | 382 |
| 48 | Diagnostic criteria for schwannomatosis. Neurology, 2005, 64, 1838-1845. | 1.1 | 368 |
| 49 | Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genetics in Medicine, 2020, 22, 15-25. | 2.4 | 365 |
| 50 | Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45. | 7.0 | 358 |
| 51 | Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691. | 21.4 | 356 |
| 52 | A clinical study of type 2 neurofibromatosis. The Quarterly Journal of Medicine, 1992, 84, 603-18. | 1.0 | 348 |
| 53 | Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102. | 27.8 | 338 |
| 54 | The effect of intermittent energy and carbohydrate restrictionv. daily energy restriction on weight loss and metabolic disease risk markers in overweight women. British Journal of Nutrition, 2013, 110, 1534-1547. | 2.3 | 336 |

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|---|---|---|---|
| 55 | 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, 140-151. | 6.2 | 335 |
| 56 | 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, 973-977. | 21.4 | 335 |
| 57 | Mortality after bilateral salpingo-oophorectomy in BRCA1 and BRCA2 mutation carriers: a prospective cohort study. Lancet Oncology, The, 2006, 7, 223-229. | 10.7 | 333 |
| 58 | 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, 93-97. | 1.3 | 328 |
| 59 | Tumor risks and genotypeÃ¢Â€Â“phenotypeÃ¢Â€Â“proteotype analysis in 358 patients with germline mutations inSDHBandSDHD. Human Mutation, 2010, 31, 41-51. | 2.5 | 325 |
| 60 | Li-Fraumeni syndrome â€“ a molecular and clinical review. British Journal of Cancer, 1997, 76, 1-14. | 6.4 | 324 |
| 61 | Critical research gaps and translational priorities for the successful prevention and treatment of breast cancer. Breast Cancer Research, 2013, 15, R92. | 5.0 | 320 |
| 62 | Second Primary Tumors in Neurofibromatosis 1 Patients Treated for Optic Glioma: Substantial Risks After Radiotherapy. Journal of Clinical Oncology, 2006, 24, 2570-2575. | 1.6 | 319 |
| 63 | Oral Contraceptives and the Risk of Breast Cancer in BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2002, 94, 1773-1779. | 6.3 | 318 |
| 64 | Germline BRCA mutation and outcome in young-onset breast cancer (POSH): a prospective cohort study. Lancet Oncology, The, 2018, 19, 169-180. | 10.7 | 316 |
| 65 | Germline E-cadherin Gene (CDH1) Mutations Predispose to Familial Gastric Cancer and Colorectal Cancer. Human Molecular Genetics, 1999, 8, 607-610. | 2.9 | 312 |
| 66 | A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892. | 21.4 | 309 |
| 67 | Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513. | 2.4 | 290 |
| 68 | The incidence of Gorlin syndrome in 173 consecutive cases of medulloblastoma. British Journal of Cancer, 1991, 64, 959-961. | 6.4 | 284 |
| 69 | Cumulative lifetime incidence of extracolonic cancers in Lynch syndrome: a report of 121 families with proven mutations. Clinical Genetics, 2009, 75, 141-149. | 2.0 | 280 |
| 70 | Effect of BRCA Mutations on Metastatic Relapse and Cause-specific Survival After Radical Treatment for Localised Prostate Cancer. European Urology, 2015, 68, 186-193. | 1.9 | 279 |
| 71 | Psychosocial impact of breast/ovarian (BRCA 1/2) cancer-predictive genetic testing in a UK multi-centre clinical cohort. British Journal of Cancer, 2004, 91, 1787-1794. | 6.4 | 276 |
| 72 | Effect of Aspirin or Resistant Starch on Colorectal Neoplasia in the Lynch Syndrome. New England Journal of Medicine, 2008, 359, 2567-2578. | 27.0 | 273 |

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|---|---|---|---|
| 73 | Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685. | 1.6 | 270 |
| 74 | Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581. | 21.4 | 265 |
| 75 | Evaluation of breast cancer risk assessment packages in the family history evaluation and screening programme. Journal of Medical Genetics, 2003, 40, 807-814. | 3.2 | 261 |
| 76 | Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. American Journal of Human Genetics, 2008, 82, 937-948. | 6.2 | 257 |
| 77 | Risk of cancer other than breast or ovarian in individuals with BRCA1 and BRCA2 mutations. Familial Cancer, 2012, 11, 235-242. | 1.9 | 252 |
| 78 | 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. | 5.0 | 252 |
| 79 | Risk determination and prevention of breast cancer. Breast Cancer Research, 2014, 16, 446. | 5.0 | 248 |
| 80 | Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212. | 3.5 | 244 |
| 81 | Are There Low-Penetrance TP53 Alleles? Evidence from Childhood Adrenocortical Tumors. American Journal of Human Genetics, 1999, 65, 995-1006. | 6.2 | 240 |
| 82 | Germline SDHD mutation in familial phaeochromocytoma. Lancet, The, 2001, 357, 1181-1182. | 13.7 | 236 |
| 83 | Germline Mutations in SUFU Cause Gorlin Syndromeâ€“Associated Childhood Medulloblastoma and Redefine the Risk Associated With PTCH1 Mutations. Journal of Clinical Oncology, 2014, 32, 4155-4161. | 1.6 | 236 |
| 84 | A new scoring system for the chances of identifying a BRCA1/2 mutation outperforms existing models including BRCAPRO. Journal of Medical Genetics, 2004, 41, 474-480. | 3.2 | 232 |
| 85 | Management of the patient and family with neurofibromatosis 2: a consensus conference statement. British Journal of Neurosurgery, 2005, 19, 5-12. | 0.8 | 229 |
| 86 | Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620. | 2.5 | 224 |
| 87 | 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. | 13.7 | 220 |
| 88 | Germline RAD51C mutations confer susceptibility to ovarian cancer. Nature Genetics, 2012, 44, 475-476. | 21.4 | 219 |
| 89 | Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. Nature, 2013, 493, 406-410. | 27.8 | 218 |
| 90 | A genetic register for von Hippel-Lindau disease.. Journal of Medical Genetics, 1996, 33, 120-127. | 3.2 | 215 |

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|---|---|---|---|
| 91 | Evaluation of clinical diagnostic criteria for neurofibromatosis 2. Neurology, 2002, 59, 1759-1765. | 1.1 | 215 |
| 92 | Predictors of the Risk of Mortality in Neurofibromatosis 2. American Journal of Human Genetics, 2002, 71, 715-723. | 6.2 | 211 |
| 93 | Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. Nature Genetics, 2013, 45, 295-298. | 21.4 | 208 |
| 94 | Prediction of pathogenic mutations in patients with early-onset breast cancer by family history. Lancet, The, 2003, 361, 1101-1102. | 13.7 | 200 |
| 95 | Germ-line mutations of TP53 in Li-Fraumeni families: an extended study of 39 families. Cancer Research, 1997, 57, 3245-52. | 0.9 | 198 |
| 96 | 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, 481-484. | 3.2 | 196 |
| 97 | Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499. | 1.9 | 195 |
| 98 | A Randomized Placebo-Controlled Prevention Trial of Aspirin and/or Resistant Starch in Young People with Familial Adenomatous Polyposis. Cancer Prevention Research, 2011, 4, 655-665. | 1.5 | 193 |
| 99 | Pregnancies, Breast-Feeding, and Breast Cancer Risk in the International BRCA1/2 Carrier Cohort Study (IBCCS). Journal of the National Cancer Institute, 2006, 98, 535-544. | 6.3 | 191 |
| 100 | Penetrance estimates for BRCA1 and BRCA2based 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. | 2.6 | 191 |
| 101 | Frequent hSNF5/INI1 Germline Mutations in Patients with Rhabdoid Tumor. Clinical Cancer Research, 2011, 17, 31-38. | 7.0 | 191 |
| 102 | EANO guideline on the diagnosis and treatment of vestibular schwannoma. Neuro-Oncology, 2020, 22, 31-45. | 1.2 | 190 |
| 103 | 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 Collaboratorsâ€™ Group. Journal of Clinical Oncology, 2006, 24, 3361-3366. | 1.6 | 188 |
| 104 | Heritability of Cellular Radiosensitivity: A Marker of Low-Penetrance Predisposition Genes in Breast Cancer?. American Journal of Human Genetics, 1999, 65, 784-794. | 6.2 | 186 |
| 105 | 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-e5660. | 6.0 | 186 |
| 106 | Cancer risk in Lynch Syndrome. Familial Cancer, 2013, 12, 229-240. | 1.9 | 186 |
| 107 | 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. | 5.0 | 186 |
| 108 | Genotype/phenotype correlations in type 2 neurofibromatosis (NF2): evidence for more severe disease associated with truncating mutations.. Journal of Medical Genetics, 1998, 35, 450-455. | 3.2 | 185 |

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|---|---|---|---|
| 109 | Cancer phenotype correlates with constitutional TP53 genotype in families with the Liâ€“Fraumeni syndrome. Oncogene, 1998, 17, 1061-1068. | 5.9 | 180 |
| 110 | Paediatric presentation of type 2 neurofibromatosis. Archives of Disease in Childhood, 1999, 81, 496-499. | 1.9 | 180 |
| 111 | Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. Journal of Medical Genetics, 2008, 45, 332-339. | 3.2 | 179 |
| 112 | Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, . | 12.6 | 178 |
| 113 | Personalized early detection and prevention of breast cancer: ENVISION consensus statement. Nature Reviews Clinical Oncology, 2020, 17, 687-705. | 27.6 | 178 |
| 114 | Predictive testing for BRCA1/2: attributes, risk perception and management in a multi-centre clinical cohort. British Journal of Cancer, 2002, 86, 1209-1216. | 6.4 | 173 |
| 115 | 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, 9742-9754. | 0.9 | 169 |
| 116 | The impact of genetic counselling on risk perception in women with a family history of breast cancer. British Journal of Cancer, 1994, 70, 934-938. | 6.4 | 168 |
| 117 | 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, 425-431. | 3.2 | 167 |
| 118 | Guidelines for the Liâ€“Fraumeni and heritable TP53-related cancer syndromes. European Journal of Human Genetics, 2020, 28, 1379-1386. | 2.8 | 167 |
| 119 | Neuroimaging manifestations in children with SARS-CoV-2 infection: a multinational, multicentre collaborative study. The Lancet Child and Adolescent Health, 2021, 5, 167-177. | 5.6 | 166 |
| 120 | Neurofibromatosis type 2. Journal of Medical Genetics, 2000, 37, 897-904. | 3.2 | 165 |
| 121 | Perception of risk in women with a family history of breast cancer. British Journal of Cancer, 1993, 67, 612-614. | 6.4 | 162 |
| 122 | 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, 1187-1191. | 2.8 | 161 |
| 123 | Bilateral Oophorectomy and Breast Cancer Risk inBRCA1andBRCA2Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, . | 6.3 | 160 |
| 124 | The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. Genetics in Medicine, 2019, 21, 2390-2400. | 2.4 | 153 |
| 125 | Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250. | 1.6 | 152 |
| 126 | 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, 5588-5596. | 1.6 | 151 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Autism and other psychiatric comorbidity in neurofibromatosis type 1: evidence from a populationâ€based study. Developmental Medicine and Child Neurology, 2013, 55, 139-145. | 2.1 | 149 |
| 128 | Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634. | 7.1 | 148 |
| 129 | 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. | 1.6 | 148 |
| 130 | Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers. European Urology, 2019, 76, 831-842. | 1.9 | 148 |
| 131 | Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95. | 27.8 | 148 |
| 132 | High resolution deletion analysis of constitutional DNA from neurofibromatosis type 2 (NF2) patients using microarray-CGH. Human Molecular Genetics, 2001, 10, 271-282. | 2.9 | 147 |
| 133 | Familial Breast Cancer. Clinical Genetics, 2012, 82, 105-114. | 2.0 | 147 |
| 134 | A genetic study of type 2 neurofibromatosis in the United Kingdom. II. Guidelines for genetic counselling.. Journal of Medical Genetics, 1992, 29, 847-852. | 3.2 | 146 |
| 135 | Neurofibromatosis Type 1 and Autism Spectrum Disorder. Pediatrics, 2013, 132, e1642-e1648. | 2.1 | 145 |
| 136 | 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, 424-428. | 3.2 | 144 |
| 137 | Contralateral mastectomy improves survival in women with BRCA1/2-associated breast cancer. Breast Cancer Research and Treatment, 2013, 140, 135-142. | 2.5 | 144 |
| 138 | Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87. | 6.2 | 144 |
| 139 | Increasing the specificity of diagnostic criteria for schwannomatosis. Neurology, 2006, 66, 730-732. | 1.1 | 143 |
| 140 | Breast cancer risk-assessment models. Breast Cancer Research, 2007, 9, 213. | 5.0 | 142 |
| 141 | Genotype-Phenotype Correlations for Nervous System Tumors in Neurofibromatosis 2: A Population-Based Study. American Journal of Human Genetics, 2004, 75, 231-239. | 6.2 | 140 |
| 142 | BRCA1, BRCA2 and TP53 mutations in very early-onset breast cancer with associated risks to relatives. European Journal of Cancer, 2006, 42, 1143-1150. | 2.8 | 139 |
| 143 | Cancer Surveillance in Gorlin Syndrome and Rhabdoid Tumor Predisposition Syndrome. Clinical Cancer Research, 2017, 23, e62-e67. | 7.0 | 139 |
| 144 | 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. | 2.4 | 139 |

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|---|---|---|---|
| 145 | Loss of SUFU Function in Familial Multiple Meningioma. American Journal of Human Genetics, 2012, 91, 520-526. | 6.2 | 137 |
| 146 | A clinical study of type 1 neurofibromatosis in north west England. Journal of Medical Genetics, 1999, 36, 197-203. | 3.2 | 137 |
| 147 | Neurofibromatosis type 1 and sporadic optic gliomas. Archives of Disease in Childhood, 2002, 87, 65-70. | 1.9 | 135 |
| 148 | Familial infiltrative fibromatosis (desmoid tumours) (MIM135290) caused by a recurrent 3' APC gene mutation. Human Molecular Genetics, 1996, 5, 1921-1924. | 2.9 | 134 |
| 149 | MRI breast screening in high-risk women: cancer detection and survival analysis. Breast Cancer Research and Treatment, 2014, 145, 663-672. | 2.5 | 133 |
| 150 | Cancer and Central Nervous System Tumor Surveillance in Pediatric Neurofibromatosis 1. Clinical Cancer Research, 2017, 23, e46-e53. | 7.0 | 133 |
| 151 | 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, 2318-2324. | 2.5 | 132 |
| 152 | Assessment of in vitro sperm characteristics in relation to fertility in dairy bulls. Animal Reproduction Science, 2008, 103, 201-214. | 1.5 | 131 |
| 153 | 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, 601-610. | 2.5 | 130 |
| 154 | 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. | 0.3 | 130 |
| 155 | Use of anastrozole for breast cancer prevention (IBIS-II): long-term results of a randomised controlled trial. Lancet, The, 2020, 395, 117-122. | 13.7 | 128 |
| 156 | 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. | 12.1 | 127 |
| 157 | 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. | 1.6 | 126 |
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