Clare Turnbull

List of Publications by Citations

105 papers	9,458 citations	46 h-index	97 g-index
122 ext. papers	11,294 ext. citations	14.1 avg, IF	5.19 L-index

#	Paper	IF	Citations
105	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
104	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
103	Genome-wide association study identifies five new breast cancer susceptibility loci. *Nature Genetics* , **2010**, 42, 504-7	36.3	582
102	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
101	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
100	Germline mutations in RAD51D confer susceptibility to ovarian cancer. *Nature Genetics*, **2011**, 43, 879-882	36.3	379
99	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
98	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
97	Variants near DMRT1, TERT and ATF7IP are associated with testicular germ cell cancer. *Nature Genetics*, **2010**, 42, 604-7	36.3	289
96	A genome-wide association study of testicular germ cell tumor. *Nature Genetics*, **2009**, 41, 807-10	36.3	282
95	DICER1 syndrome: clarifying the diagnosis, clinical features and management implications of a pleiotropic tumour predisposition syndrome. *Journal of Medical Genetics*, **2011**, 48, 273-8	5.8	278
94	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
93	Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases: A Mendelian Randomization Study. *JAMA Oncology*, **2017**, 3, 636-651	13.4	236
92	Genetic predisposition to breast cancer: past, present, and future. *Annual Review of Genomics and Human Genetics*, **2008**, 9, 321-45	9.7	202
91	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. *Nature*, **2013**, 493, 406-10	50.4	191
90	Germline RAD51C mutations confer susceptibility to ovarian cancer. *Nature Genetics*, **2012**, 44, 475-6; author reply 476	36.3	190
89	The 100 000 Genomes Project: bringing whole genome sequencing to the NHS. *BMJ, The*, **2018**, 361, k1687	5.9	184

88	Effect of delays in the 2-week-wait cancer referral pathway during the COVID-19 pandemic on cancer survival in the UK: a modelling study. *Lancet Oncology, The*, **2020**, 21, 1035-1044	21.7	184
87	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. *Human Molecular Genetics*, **2012**, 21, 5373-84	5.6	143
86	Meta-analysis identifies four new loci associated with testicular germ cell tumor. *Nature Genetics*, **2013**, 45, 680-5	36.3	132
85	Whole-exome sequencing reveals the mutational spectrum of testicular germ cell tumours. *Nature Communications*, **2015**, 6, 5973	17.4	128
84	Identification of nine new susceptibility loci for testicular cancer, including variants near DAZL and PRDM14. *Nature Genetics*, **2013**, 45, 686-9	36.3	128
83	Genomic evolution and chemoresistance in germ-cell tumours. *Nature*, **2016**, 540, 114-118	50.4	100
82	Estimated impact of the COVID-19 pandemic on cancer services and excess 1-year mortality in people with cancer and multimorbidity: near real-time data on cancer care, cancer deaths and a population-based cohort study. *BMJ Open*, **2020**, 10, e043828	3	96
81	Cost-effectiveness of Population-Based BRCA1, BRCA2, RAD51C, RAD51D, BRIP1, PALB2 Mutation Testing in Unselected General Population Women. *Journal of the National Cancer Institute*, **2018**, 110, 714-725	9.7	92
80	Risk of estrogen receptor-positive and -negative breast cancer and single-nucleotide polymorphism 2q35-rs13387042. *Journal of the National Cancer Institute*, **2009**, 101, 1012-8	9.7	90
79	Identification of 19 new risk loci and potential regulatory mechanisms influencing susceptibility to testicular germ cell tumor. *Nature Genetics*, **2017**, 49, 1133-1140	36.3	89
78	Meta-analysis of five genome-wide association studies identifies multiple new loci associated with testicular germ cell tumor. *Nature Genetics*, **2017**, 49, 1141-1147	36.3	85
77	Genetic predisposition to mosaic Y chromosome loss in blood. *Nature*, **2019**, 575, 652-657	50.4	83
76	Stratification of Wilms tumor by genetic and epigenetic analysis. *Oncotarget*, **2012**, 3, 327-35	3.3	78
75	Clinical whole-genome sequencing from routine formalin-fixed, paraffin-embedded specimens: pilot study for the 100,000 Genomes Project. *Genetics in Medicine*, **2018**, 20, 1196-1205	8.1	77
74	Population genetic testing for cancer susceptibility: founder mutations to genomes. *Nature Reviews Clinical Oncology*, **2016**, 13, 41-54	19.4	67
73	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
72	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. *Cancer Research*, **2016**, 76, 5103-14	10.1	66
71	The genomic landscape of testicular germ cell tumours: from susceptibility to treatment. *Nature Reviews Urology*, **2016**, 13, 409-19	5.5	65

70	Germline-focussed analysis of tumour-only sequencing: recommendations from the ESMO Precision Medicine Working Group. *Annals of Oncology*, **2019**, 30, 1221-1231	10.3	64
69	Personalized early detection and prevention of breast cancer: ENVISION consensus statement. *Nature Reviews Clinical Oncology*, **2020**, 17, 687-705	19.4	64
68	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
67	A genome-wide association study of early-onset breast cancer identifies PFKM as a novel breast cancer gene and supports a common genetic spectrum for breast cancer at any age. *Cancer Epidemiology Biomarkers and Prevention*, **2014**, 23, 658-69	4	63
66	A genome-wide association study identifies susceptibility loci for Wilms tumor. *Nature Genetics*, **2012**, 44, 681-4	36.3	58
65	Cancer genetics, precision prevention and a call to action. *Nature Genetics*, **2018**, 50, 1212-1218	36.3	57
64	Consensus for genes to be included on cancer panel tests offered by UK genetics services: guidelines of the UK Cancer Genetics Group. *Journal of Medical Genetics*, **2018**, 55, 372-377	5.8	55
63	Testicular germ cell tumor susceptibility associated with the UCK2 locus on chromosome 1q23. *Human Molecular Genetics*, **2013**, 22, 2748-53	5.6	53
62	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. *Journal of the National Cancer Institute*, **2020**, 112, 1242-1250	9.7	51
61	Mammographic breast density and breast cancer: evidence of a shared genetic basis. *Cancer Research*, **2012**, 72, 1478-84	10.1	50
60	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
59	Predisposition gene identification in common cancers by exome sequencing: insights from familial breast cancer. *Breast Cancer Research and Treatment*, **2012**, 134, 429-33	4.4	46
58	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
57	Large-scale Sequencing of Testicular Germ Cell Tumour (TGCT) Cases Excludes Major TGCT Predisposition Gene. *European Urology*, **2018**, 73, 828-831	10.2	38
56	Quantifying the heritability of testicular germ cell tumour using both population-based and genomic approaches. *Scientific Reports*, **2015**, 5, 13889	4.9	38
55	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
54	Two new loci and gene sets related to sex determination and cancer progression are associated with susceptibility to testicular germ cell tumor. *Human Molecular Genetics*, **2015**, 24, 4138-46	5.6	36
53	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35

52	Gene-gene interactions in breast cancer susceptibility. *Human Molecular Genetics*, **2012**, 21, 958-62	5.6	33
51	Assessment of polygenic architecture and risk prediction based on common variants across fourteen cancers. *Nature Communications*, **2020**, 11, 3353	17.4	32
50	Identification of four new susceptibility loci for testicular germ cell tumour. *Nature Communications* , **2015**, 6, 8690	17.4	30
49	Current detection rates and time-to-detection of all identifiable carriers in the Greater London population. *Journal of Medical Genetics*, **2018**, 55, 538-545	5.8	30
48	Development of cancer genetic services in the UK: A national consultation. *Genome Medicine*, **2015**, 7, 18	14.4	29
47	Genome-wide association study of germline variants and breast cancer-specific mortality. *British Journal of Cancer*, **2019**, 120, 647-657	8.7	28
46	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
45	Multi-stage genome-wide association study identifies new susceptibility locus for testicular germ cell tumour on chromosome 3q25. *Human Molecular Genetics*, **2015**, 24, 1169-76	5.6	26
44	Genetic predisposition to cancer. *Clinical Medicine*, **2005**, 5, 491-8	1.9	26
43	Rare disruptive mutations in ciliary function genes contribute to testicular cancer susceptibility. *Nature Communications*, **2016**, 7, 13840	17.4	26
42	Pathway-based analysis of GWAs data identifies association of sex determination genes with susceptibility to testicular germ cell tumors. *Human Molecular Genetics*, **2014**, 23, 6061-8	5.6	25
41	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. *Breast Cancer Research*, **2016**, 18, 64	8.3	25
40	p.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancer. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 4275-4282	5.6	24
39	Genomic landscape of platinum resistant and sensitive testicular cancers. *Nature Communications*, **2020**, 11, 2189	17.4	23
38	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
37	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
36	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. *Journal of Medical Genetics*, **2019**, 56, 347-357	5.8	19
35	Clinical practice guidelines for BRCA1 and BRCA2 genetic testing. *European Journal of Cancer*, **2021**, 146, 30-47	7.5	15

34	Large-scale Analysis Demonstrates Familial Testicular Cancer to have Polygenic Aetiology. *European Urology*, **2018**, 74, 248-252	10.2	13
33	Mendelian randomisation study of the relationship between vitamin D and risk of glioma. *Scientific Reports*, **2018**, 8, 2339	4.9	12
32	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. *Journal of Medical Genetics*, **2020**, 57, 829-834	5.8	11
31	Mendelian randomization provides support for obesity as a risk factor for meningioma. *Scientific Reports*, **2019**, 9, 309	4.9	10
30	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
29	Association study of prostate cancer susceptibility variants with risks of invasive ovarian, breast, and colorectal cancer. *Cancer Research*, **2008**, 68, 8837-42	10.1	10
28	Polygenic susceptibility to testicular cancer: implications for personalised health care. *British Journal of Cancer*, **2015**, 113, 1512-8	8.7	9
27	Collateral damage: the impact on cancer outcomes of the COVID-19 pandemic		9
26	Etiologic Index - A Case-Only Measure of -Associated Cancer Risk. *New England Journal of Medicine*, **2020**, 383, 286-288	59.2	9
25	Testicular cancer in 2017: Sequencing advances understanding. *Nature Reviews Urology*, **2018**, 15, 79-80	5.5	8
24	Subphenotype meta-analysis of testicular cancer genome-wide association study data suggests a role for RBFOX family genes in cryptorchidism susceptibility. *Human Reproduction*, **2018**, 33, 967-977	5.7	7
23	Mutation and association analysis of GEN1 in breast cancer susceptibility. *Breast Cancer Research and Treatment*, **2010**, 124, 283-8	4.4	7
22	Combining evidence for and against pathogenicity for variants in cancer susceptibility genes: CanVIG-UK consensus recommendations. *Journal of Medical Genetics*, **2021**, 58, 297-304	5.8	7
21	Germline and Somatic Genetic Variants in the p53 Pathway Interact to Affect Cancer Risk, Progression, and Drug Response. *Cancer Research*, **2021**, 81, 1667-1680	10.1	7
20	Validation of loci at 2q14.2 and 15q21.3 as risk factors for testicular cancer. *Oncotarget*, **2018**, 9, 12630-12638	5.5	6
19	Concern regarding classification of germline TP53 variants as likely pathogenic. *Human Mutation*, **2019**, 40, 828-831	4.7	5
18	Primrose syndrome: Characterization of the phenotype in 42 patients. *Clinical Genetics*, **2020**, 97, 890-901	4	5
17	Genetic predisposition to mosaic Y chromosome loss in blood is associated with genomic instability in other tissues and susceptibility to non-haematological cancers		5

16	Clinical likelihood ratios and balanced accuracy for 44 in silico tools against multiple large-scale functional assays of cancer susceptibility genes. *Genetics in Medicine*, **2021**, 23, 2096-2104	8.1	5
15	Identification of 22 susceptibility loci associated with testicular germ cell tumors. *Nature Communications*, **2021**, 12, 4487	17.4	5
14	Effect of COVID-19 on colorectal cancer care in England. *The Lancet Gastroenterology and Hepatology*, **2021**, 6, 152-154	18.8	4
13	Structural Aberrations with Secondary Implications (SASIs): consensus recommendations for reporting of cancer susceptibility genes identified during analysis of Copy Number Variants (CNVs). *Journal of Medical Genetics*, **2019**, 56, 718-726	5.8	3
12	Tumor-only sequencing for oncology management: Germline-focused analysis and implications. *Genes Chromosomes and Cancer*, **2021**, 60, 352-357	5	3
11	Response to Letter to the Editor: "p.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancer". *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 3518-3519	5.6	3
10	Nasopharyngeal teratoma and diaphragmatic hernia: no longer a random association but a new syndrome?. *Clinical Dysmorphology*, **2009**, 18, 131-134	0.9	2
9	Germline and somatic genetic variants in the p53 pathway interact to affect cancer risk, progression and drug response		2
8	Quantifying and mitigating the impact of the COVID-19 pandemic on outcomes in colorectal cancer		1
7	Assessment of Polygenic Architecture and Risk Prediction based on Common Variants Across Fourteen Cancers		1
6	Rapid reversal of clinical down-classification of a BRCA1 splicing variant avoiding psychological harm. *Clinical Genetics*, **2019**, 95, 532-533	4	1
5	Genomics in medicine. *Medicine*, **2018**, 46, 774-779	0.6	1
4	Peridiagnostic and cascade cancer genetic testing. *Nature Reviews Clinical Oncology*, **2020**, 17, 277-278	19.4	0
3	Genetically Inferred Telomere Length and Testicular Germ Cell Tumor Risk. *Cancer Epidemiology Biomarkers and Prevention*, **2021**, 30, 1275-1278	4	0
2	Screening for testicular cancer **2018**, 349-360		
1	Phenotype evaluation and clinical context: application of case-level data in genomic variant interpretation **2021**, 251-274		