

Katherine L Nathanson

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

343
papers

25,504
citations

81
h-index

153
g-index

363
ext. papers

30,522
ext. citations

10.1
avg, IF

6.39
L-index

#	Paper	IF	Citations
343	Evaluation of Classic, Attenuated, and Oligopolyposis of the Colon. <i>Gastrointestinal Endoscopy Clinics of North America</i> , 2022 , 32, 95-112	3.3	
342	Abstract OT2-18-01: Harnessing olaparib, palbociclib, and endocrine therapy (HOPE): Phase I/II trial of olaparib, palbociclib and fulvestrant in patients with BRCA1/2-associated, hormone receptor-positive, HER2-negative metastatic breast cancer. <i>Cancer Research</i> , 2022 , 82, OT2-18-01-OT2-18-01	10.1	
341	Breast Cancer Screening Strategies for Women With ATM, CHEK2, and PALB2 Pathogenic Variants: A Comparative Modeling Analysis.. <i>JAMA Oncology</i> , 2022 ,	13.4	5
340	Abstract P2-09-01: Population-based risk estimates of clinical subtypes of breast cancer among carriers of germline pathogenic variants in cancer predisposition genes. <i>Cancer Research</i> , 2022 , 82, P2-09-01-P2-09-01	10.1	
339	Loss and Promoter Hypermethylation Negatively Predict for Immunogenicity in BRCA-Deficient Ovarian Cancer.. <i>JCO Precision Oncology</i> , 2022 , 6, e2100159	3.6	0
338	Performance of polygenic risk scores for cancer prediction in a racially diverse academic biobank.. <i>Genetics in Medicine</i> , 2021 ,	8.1	2
337	Uptake and outcomes of small intestinal and urinary tract cancer surveillance in Lynch syndrome.. <i>World Journal of Clinical Oncology</i> , 2021 , 12, 1023-1036	2.5	
336	Targeted BRCA1/2 population screening among Ashkenazi Jewish individuals using a web-enabled medical model: An observational cohort study.. <i>Genetics in Medicine</i> , 2021 ,	8.1	1
335	Germline Pathogenic Variants in Cancer Predisposition Genes Among Women With Invasive Lobular Carcinoma of the Breast. <i>Journal of Clinical Oncology</i> , 2021 , 39, 3918-3926	2.2	6
334	Evaluating Polygenic Risk Scores for Breast Cancer in Women of African Ancestry. <i>Journal of the National Cancer Institute</i> , 2021 , 113, 1168-1176	9.7	9
333	Discovery and fine-mapping of height loci via high-density imputation of GWASs in individuals of African ancestry. <i>American Journal of Human Genetics</i> , 2021 , 108, 564-582	11	7
332	HLA Class I Polymorphisms Influencing Both Peptide Binding and KIR Interactions Are Associated with Remission among Children with Atopic Dermatitis: A Longitudinal Study. <i>Journal of Immunology</i> , 2021 , 206, 2038-2044	5.3	3
331	Multigene Panel Testing in Individuals With Hepatocellular Carcinoma Identifies Pathogenic Germline Variants. <i>JCO Precision Oncology</i> , 2021 , 5,	3.6	2
330	Evolution of delayed resistance to immunotherapy in a melanoma responder. <i>Nature Medicine</i> , 2021 , 27, 985-992	50.5	11
329	Mastermind Like Transcriptional Coactivator 3 (MAML3) Drives Neuroendocrine Tumor Progression. <i>Molecular Cancer Research</i> , 2021 , 19, 1476-1485	6.6	3
328	Challenges and Opportunities in Engaging Primary Care Providers in BRCA Testing: Results from the BFOR Study. <i>Journal of General Internal Medicine</i> , 2021 , 1	4	2
327	Correlation Between Plasma Catecholamines, Weight, and Diabetes in Pheochromocytoma and Paraganglioma. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e4028-e4038	5.6	6

326	Germline POT1 variants can predispose to myeloid and lymphoid neoplasms. <i>Leukemia</i> , 2021 ,	10.7	2
325	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. <i>Genetics in Medicine</i> , 2021 , 23, 1726-1737	8.1	2
324	Cross-ancestry GWAS meta-analysis identifies six breast cancer loci in African and European ancestry women. <i>Nature Communications</i> , 2021 , 12, 4198	17.4	1
323	Identification of 22 susceptibility loci associated with testicular germ cell tumors. <i>Nature Communications</i> , 2021 , 12, 4487	17.4	5
322	Risk of Late-Onset Breast Cancer in Genetically Predisposed Women. <i>Journal of Clinical Oncology</i> , 2021 , 39, 3430-3440	2.2	3
321	TSLP and IL-7R Variants Are Associated with Persistent Atopic Dermatitis. <i>Journal of Investigative Dermatology</i> , 2021 , 141, 446-450.e2	4.3	5
320	Real-world integration of genomic data into the electronic health record: the PennChart Genomics Initiative. <i>Genetics in Medicine</i> , 2021 , 23, 603-605	8.1	5
319	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , 2021 , 12, 1078	17.4	4
318	From Race-Based to Precision Oncology: Leveraging Behavioral Economics and the Electronic Health Record to Advance Health Equity in Cancer Care. <i>JCO Precision Oncology</i> , 2021 , 5,	3.6	2
317	A Population-Based Study of Genes Previously Implicated in Breast Cancer. <i>New England Journal of Medicine</i> , 2021 , 384, 440-451	59.2	115
316	Clinical Management of Oligopolyposis of Unknown Etiology. <i>Current Treatment Options in Gastroenterology</i> , 2021 , 19, 183-197	2.5	1
315	Genetically Inferred Telomere Length and Testicular Germ Cell Tumor Risk. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2021 , 30, 1275-1278	4	0
314	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2021 ,	9.7	3
313	Comparison of the Prevalence of Pathogenic Variants in Cancer Susceptibility Genes in Black Women and Non-Hispanic White Women With Breast Cancer in the United States. <i>JAMA Oncology</i> , 2021 , 7, 1045-1050	13.4	7
312	Genetic risk assessment for hereditary renal cell carcinoma: Clinical consensus statement. <i>Cancer</i> , 2021 , 127, 3957-3966	6.4	1
311	EUS-based Pancreatic Cancer Surveillance in Carriers Without a Family History of Pancreatic Cancer. <i>Cancer Prevention Research</i> , 2021 , 14, 1033-1040	3.2	0
310	Using a Machine Learning Approach to Identify Low-Frequency and Rare Alleles Associated with Remission of Atopic Dermatitis.. <i>JID Innovations</i> , 2021 , 1, 100046		0
309	Risk of Breast Cancer Among Carriers of Pathogenic Variants in Breast Cancer Predisposition Genes Varies by Polygenic Risk Score. <i>Journal of Clinical Oncology</i> , 2021 , 39, 2564-2573	2.2	12

308	Phase II Study of Maintenance Rucaparib in Patients With Platinum-Sensitive Advanced Pancreatic Cancer and a Pathogenic Germline or Somatic Variant in , , or. <i>Journal of Clinical Oncology</i> , 2021 , 39, 2497-2505 ³⁴		
307	copy number is a biomarker for response to combination WEE1-ATR inhibition in ovarian and endometrial cancer models. <i>Cell Reports Medicine</i> , 2021 , 2, 100394	18	2
306	Targeting PHGDH Upregulation Reduces Glutathione Levels and Resensitizes Resistant NRAS-Mutant Melanoma to MAPK Kinase Inhibition. <i>Journal of Investigative Dermatology</i> , 2020 , 140, 2242-2252.e7	4.3	11
305	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020 , 52, 572-581	36.3	76
304	Contribution of Germline Predisposition Gene Mutations to Breast Cancer Risk in African American Women. <i>Journal of the National Cancer Institute</i> , 2020 , 112, 1213-1221	9.7	25
303	Points to consider for reporting of germline variation in patients undergoing tumor testing: a statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2020 , 22, 1142-1148	8.1	35
302	Longitudinal follow-up after telephone disclosure in the randomized COGENT study. <i>Genetics in Medicine</i> , 2020 , 22, 1401-1406	8.1	1
301	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). <i>JAMA Oncology</i> , 2020 , 6, 1218-1230	13.4	25
300	Associations of sociodemographic and clinical factors with gastrointestinal cancer risk assessment appointment completion. <i>Journal of Genetic Counseling</i> , 2020 , 29, 616-624	2.5	0
299	Assessment of polygenic architecture and risk prediction based on common variants across fourteen cancers. <i>Nature Communications</i> , 2020 , 11, 3353	17.4	32
298	Longitudinal outcomes with cancer multigene panel testing in previously tested BRCA1/2 negative patients. <i>Clinical Genetics</i> , 2020 , 97, 601-609	4	3
297	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. <i>Genetic Epidemiology</i> , 2020 , 44, 442-468	2.6	9
296	Frequency of radiation-induced malignancies post-adjuvant radiotherapy for breast cancer in patients with Li-Fraumeni syndrome. <i>Breast Cancer Research and Treatment</i> , 2020 , 181, 181-188	4.4	14
295	Association of premenopausal risk-reducing salpingo-oophorectomy with breast cancer risk in BRCA1/2 mutation carriers: Maximising bias-reduction. <i>European Journal of Cancer</i> , 2020 , 132, 53-60	7.5	10
294	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020 , 80, 624-638	10.1	22
293	Germline POT1 Variants Can Predispose to a Variety of Hematologic Neoplasms. <i>Blood</i> , 2020 , 136, 2-4	2.2	
292	Lower abdominal and pelvic radiation and testicular germ cell tumor risk. <i>PLoS ONE</i> , 2020 , 15, e0239321 ^{3,7}		1
291	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020 , 52, 56-73	36.3	56

290	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020 , 38, 674-685	2.2	133
289	Platinum response characteristics of patients with pancreatic ductal adenocarcinoma and a germline BRCA1, BRCA2 or PALB2 mutation. <i>British Journal of Cancer</i> , 2020 , 122, 333-339	8.7	60
288	Endoscopic Ultrasound Has Limited Utility in Diagnosis of Gastric Cancer in Carriers of CDH1 Mutations. <i>Clinical Gastroenterology and Hepatology</i> , 2020 , 18, 505-508.e1	6.9	12
287	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , 2020 , 22, 1653-1666	8.1	34
286	A Rare Mutation Predominant in Ashkenazi Jews Confers Risk of Multiple Cancers. <i>Cancer Research</i> , 2020 , 80, 3732-3744	10.1	7
285	Lack of pathogenic germline DICER1 variants in males with testicular germ-cell tumors. <i>Cancer Genetics</i> , 2020 , 248-249, 49-56	2.3	
284	Tumor detection rates in screening of individuals with SDHx-related hereditary paraganglioma-pheochromocytoma syndrome. <i>Genetics in Medicine</i> , 2020 , 22, 2101-2107	8.1	9
283	Predicting Metastatic Potential in Pheochromocytoma and Paraganglioma: A Comparison of PASS and GAPP Scoring Systems. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020 , 105,	5.6	10
282	Upper Endoscopic Surveillance in Lynch Syndrome Detects Gastric and Duodenal Adenocarcinomas. <i>Cancer Prevention Research</i> , 2020 , 13, 1047-1054	3.2	13
281	Association of germline variation with the survival of women with pathogenic variants and breast cancer. <i>Npj Breast Cancer</i> , 2020 , 6, 44	7.8	3
280	Mutation Rates in Cancer Susceptibility Genes in Patients With Breast Cancer With Multiple Primary Cancers. <i>JCO Precision Oncology</i> , 2020 , 4,	3.6	2
279	Upper Gastrointestinal Cancer Risk and Surveillance Outcomes in Li-Fraumeni Syndrome. <i>American Journal of Gastroenterology</i> , 2020 , 115, 2095-2097	0.7	3
278	Filaggrin sequencing and bioinformatics tools. <i>Archives of Dermatological Research</i> , 2020 , 312, 155-158	3.3	6
277	Association between fine mapping thymic stromal lymphopoietin and atopic dermatitis onset and persistence. <i>Annals of Allergy, Asthma and Immunology</i> , 2019 , 123, 595-601.e1	3.2	6
276	Hi-Plex2: a simple and robust approach to targeted sequencing-based genetic screening. <i>BioTechniques</i> , 2019 , 67, 118-122	2.5	6
275	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019 , 10, 431	17.4	45
274	Association of Inherited Pathogenic Variants in Checkpoint Kinase 2 (CHEK2) With Susceptibility to Testicular Germ Cell Tumors. <i>JAMA Oncology</i> , 2019 , 5, 514-522	13.4	23
273	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019 , 40, 1557-1578	4.7	52

272	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , 2019 , 121, 180-192	8.7	13
271	BRCA1 and BRCA2 pathogenic sequence variants in women of African origin or ancestry. <i>Human Mutation</i> , 2019 , 40, 1781-1796	4.7	16
270	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019 , 10, 1741	17.4	47
269	Genomic Signatures Predict the Immunogenicity of BRCA-Deficient Breast Cancer. <i>Clinical Cancer Research</i> , 2019 , 25, 4363-4374	12.9	38
268	A single dose of neoadjuvant PD-1 blockade predicts clinical outcomes in resectable melanoma. <i>Nature Medicine</i> , 2019 , 25, 454-461	50.5	283
267	Opposing Functions of Interferon Coordinate Adaptive and Innate Immune Responses to Cancer Immune Checkpoint Blockade. <i>Cell</i> , 2019 , 178, 933-948.e14	56.2	141
266	Association of Filaggrin Loss-of-Function Variants With Race in Children With Atopic Dermatitis. <i>JAMA Dermatology</i> , 2019 , 155, 1269-1276	5.1	16
265	The role of endoscopy in the management of hereditary diffuse gastric cancer syndrome. <i>World Journal of Gastroenterology</i> , 2019 , 25, 2878-2886	5.6	22
264	ERK1/2 Reporter Predictively Models Response and Resistance to Combined BRAF and MEK Inhibitors in Melanoma. <i>Molecular Cancer Therapeutics</i> , 2019 , 18, 1637-1648	6.1	7
263	Research participants' experiences with return of genetic research results and preferences for web-based alternatives. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e898	2.3	11
262	The p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019 , 5, 38	7.8	12
261	Identification and Confirmation of Potentially Actionable Germline Mutations in Tumor-Only Genomic Sequencing. <i>JCO Precision Oncology</i> , 2019 , 3,	3.6	9
260	Preferences for in-person disclosure: Patients declining telephone disclosure characteristics and outcomes in the multicenter Communication Of GENetic Test Results by Telephone study. <i>Clinical Genetics</i> , 2019 , 95, 293-301	4	8
259	Earlier Colorectal Cancer Screening May Be Necessary In Patients With Li-Fraumeni Syndrome. <i>Gastroenterology</i> , 2019 , 156, 273-274	13.3	14
258	NRAS Q61R and BRAF G466A mutations in atypical melanocytic lesions newly arising in advanced melanoma patients treated with vemurafenib. <i>Journal of Cutaneous Pathology</i> , 2019 , 46, 190-194	1.7	3
257	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019 , 111, 350-364	9.7	22
256	Response to Hannah-Shmouni and Stratakis. <i>Genetics in Medicine</i> , 2019 , 21, 1256	8.1	
255	Randomized Noninferiority Trial of Telephone vs In-Person Disclosure of Germline Cancer Genetic Test Results. <i>Journal of the National Cancer Institute</i> , 2018 , 110, 985-993	9.7	25

254	Clinical germline diagnostic exome sequencing for hereditary cancer: Findings within novel candidate genes are prevalent. <i>Cancer Genetics</i> , 2018 , 224-225, 12-20	2.3	4
253	Subphenotype meta-analysis of testicular cancer genome-wide association study data suggests a role for RBFOX family genes in cryptorchidism susceptibility. <i>Human Reproduction</i> , 2018 , 33, 967-977	5.7	7
252	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , 2018 , 39, 593-620	4.7	138
251	Uncommon Filaggrin Variants Are Associated with Persistent Atopic Dermatitis in African Americans. <i>Journal of Investigative Dermatology</i> , 2018 , 138, 1501-1506	4.3	39
250	Tumor Immunity and Survival as a Function of Alternative Neopeptides in Human Cancer. <i>Cancer Immunology Research</i> , 2018 , 6, 276-287	12.5	46
249	Association of Pancreatic Cancer Susceptibility Variants with Risk of Breast Cancer in Women of European and African Ancestry. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2018 , 27, 116-118	4	2
248	Genetic variants demonstrating flip-flop phenomenon and breast cancer risk prediction among women of African ancestry. <i>Breast Cancer Research and Treatment</i> , 2018 , 168, 703-712	4.4	22
247	Care of adults with neurofibromatosis type 1: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2018 , 20, 671-682	8.1	74
246	Induction of Telomere Dysfunction Prolongs Disease Control of Therapy-Resistant Melanoma. <i>Clinical Cancer Research</i> , 2018 , 24, 4771-4784	12.9	21
245	Genetic variation in the vitamin D related pathway and breast cancer risk in women of African ancestry in the root consortium. <i>International Journal of Cancer</i> , 2018 , 142, 36-43	7.5	7
244	Arginase 2 Suppresses Renal Carcinoma Progression via Biosynthetic Cofactor Pyridoxal Phosphate Depletion and Increased Polyamine Toxicity. <i>Cell Metabolism</i> , 2018 , 27, 1263-1280.e6	24.6	45
243	Genetic variation in the Hippo pathway and breast cancer risk in women of African ancestry. <i>Molecular Carcinogenesis</i> , 2018 , 57, 1311-1318	5	6
242	Integrated Molecular Characterization of Testicular Germ Cell Tumors. <i>Cell Reports</i> , 2018 , 23, 3392-3406	10.6	200
241	Retrospective Survival Analysis of Patients With Advanced Pancreatic Ductal Adenocarcinoma and Germline or Mutations.. <i>JCO Precision Oncology</i> , 2018 , 2, 1-9	3.6	11
240	Returning Individual Genetic Research Results to Research Participants: Uptake and Outcomes Among Patients With Breast Cancer. <i>JCO Precision Oncology</i> , 2018 , 2,	3.6	8
239	Use and Patient-Reported Outcomes of Clinical Multigene Panel Testing for Cancer Susceptibility in the Multicenter Communication of Genetic Test Results by Telephone Study. <i>JCO Precision Oncology</i> , 2018 , 2,	3.6	8
238	CODEX2: full-spectrum copy number variation detection by high-throughput DNA sequencing. <i>Genome Biology</i> , 2018 , 19, 202	18.3	34
237	A practical approach to adjusting for population stratification in genome-wide association studies: principal components and propensity scores (PCAPS). <i>Statistical Applications in Genetics and Molecular Biology</i> , 2018 , 17,	1.2	11

236	Comprehensive Molecular Characterization of Pheochromocytoma and Paraganglioma. <i>Cancer Cell</i> , 2017 , 31, 181-193	24.3	350
235	Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for BRCA1 pathogenic variant carriers. <i>European Journal of Human Genetics</i> , 2017 , 25, 432-438	5.3	15
234	T-cell invigoration to tumour burden ratio associated with anti-PD-1 response. <i>Nature</i> , 2017 , 545, 60-65	50.4	850
233	Application of Panel-Based Tests for Inherited Risk of Cancer. <i>Annual Review of Genomics and Human Genetics</i> , 2017 , 18, 201-227	9.7	23
232	Functional characterization of a multi-cancer risk locus on chr5p15.33 reveals regulation of TERT by ZNF148. <i>Nature Communications</i> , 2017 , 8, 15034	17.4	26
231	Von Hippel-Lindau and Hereditary Pheochromocytoma/Paraganglioma Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. <i>Clinical Cancer Research</i> , 2017 , 23, e68-e75	12.9	127
230	Recommendations for Cancer Surveillance in Individuals with RASopathies and Other Rare Genetic Conditions with Increased Cancer Risk. <i>Clinical Cancer Research</i> , 2017 , 23, e83-e90	12.9	77
229	The contribution of pathogenic variants in breast cancer susceptibility genes to familial breast cancer risk. <i>Npj Breast Cancer</i> , 2017 , 3, 22	7.8	78
228	Rare cell variability and drug-induced reprogramming as a mode of cancer drug resistance. <i>Nature</i> , 2017 , 546, 431-435	50.4	544
227	A practical guide for evaluating gonadal germ cell tumor predisposition in differences of sex development. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017 , 175, 304-314	31.4	29
226	Meta-analysis of five genome-wide association studies identifies multiple new loci associated with testicular germ cell tumor. <i>Nature Genetics</i> , 2017 , 49, 1141-1147	36.3	85
225	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017 , 49, 680-691	36.3	190
224	Characterizing Genetic Susceptibility to Breast Cancer in Women of African Ancestry. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2017 , 26, 1016-1026	4	12
223	Pheochromocytoma and Paraganglioma Susceptibility Genes: Estimating the Associated Risk of Disease. <i>JAMA Oncology</i> , 2017 , 3, 1212-1213	13.4	2
222	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017 , 49, 1767-1778	36.3	186
221	Reply to R.L. Nussbaum et al and J.S. Dolinsky et al. <i>Journal of Clinical Oncology</i> , 2017 , 35, 1262-1263	2.2	1
220	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017 , 35, 2240-2250	2.2	101
219	BRCA locus-specific loss of heterozygosity in germline BRCA1 and BRCA2 carriers. <i>Nature Communications</i> , 2017 , 8, 319	17.4	139

218	ALLELE-SPECIFIC COPY NUMBER ESTIMATION BY WHOLE EXOME SEQUENCING. <i>Annals of Applied Statistics</i> , 2017 , 11, 1169-1192	2.1	6
217	Association of breast cancer risk and the mTOR pathway in women of African ancestry in The RootQ Consortium. <i>Carcinogenesis</i> , 2017 , 38, 789-796	4.6	3
216	Genetic and Genomic Characterization of 462 Melanoma Patient-Derived Xenografts, Tumor Biopsies, and Cell Lines. <i>Cell Reports</i> , 2017 , 21, 1936-1952	10.6	45
215	A Comprehensive Patient-Derived Xenograft Collection Representing the Heterogeneity of Melanoma. <i>Cell Reports</i> , 2017 , 21, 1953-1967	10.6	89
214	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. <i>Clinical Cancer Research</i> , 2017 , 23, e107-e114	12.9	62
213	Multiple Endocrine Neoplasia and Hyperparathyroid-Jaw Tumor Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. <i>Clinical Cancer Research</i> , 2017 , 23, e123-e132	12.9	43
212	A functionally significant SNP in TP53 and breast cancer risk in African-American women. <i>Npj Breast Cancer</i> , 2017 , 3, 5	7.8	29
211	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. <i>Breast Cancer Research and Treatment</i> , 2017 , 161, 117-134	4.4	15
210	A patient-derived-xenograft platform to study BRCA-deficient ovarian cancers. <i>JCI Insight</i> , 2017 , 2, e89760	6.9	49
209	Discovery and fine-mapping of adiposity loci using high density imputation of genome-wide association studies in individuals of African ancestry: African Ancestry Anthropometry Genetics Consortium. <i>PLoS Genetics</i> , 2017 , 13, e1006719	6	60
208	Association of breast cancer risk in women of African ancestry with genetic variants in the TET-related DNA demethylation pathway.. <i>Journal of Clinical Oncology</i> , 2017 , 35, e13015-e13015	2.2	
207	Copy Number Changes Are Associated with Response to Treatment with Carboplatin, Paclitaxel, and Sorafenib in Melanoma. <i>Clinical Cancer Research</i> , 2016 , 22, 374-82	12.9	28
206	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016 , 141, 386-401	4.9	15
205	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. <i>Breast Cancer Research</i> , 2016 , 18, 112	8.3	25
204	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016 , 7, 11375	17.4	64
203	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016 , 7, 12675	17.4	53
202	Counselling framework for moderate-penetrance cancer-susceptibility mutations. <i>Nature Reviews Clinical Oncology</i> , 2016 , 13, 581-8	19.4	200
201	Patient feedback and early outcome data with a novel tiered-binned model for multiplex breast cancer susceptibility testing. <i>Genetics in Medicine</i> , 2016 , 18, 25-33	8.1	44

200	Genomic Biomarkers for Breast Cancer Risk. <i>Advances in Experimental Medicine and Biology</i> , 2016 , 882, 1-32	3.6	26
199	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016 , 48, 374-86	36.3	93
198	Personalized Preclinical Trials in BRAF Inhibitor-Resistant Patient-Derived Xenograft Models Identify Second-Line Combination Therapies. <i>Clinical Cancer Research</i> , 2016 , 22, 1592-602	12.9	91
197	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016 , 108,	9.7	65
196	Muscle oxidative phosphorylation quantitation using creatine chemical exchange saturation transfer (CrCEST) MRI in mitochondrial disorders. <i>JCI Insight</i> , 2016 , 1, e88207	9.9	26
195	PIM kinases as therapeutic targets against advanced melanoma. <i>Oncotarget</i> , 2016 , 7, 54897-54912	3.3	14
194	Targeting Notch enhances the efficacy of ERK inhibitors in BRAF-V600E melanoma. <i>Oncotarget</i> , 2016 , 7, 71211-71222	3.3	9
193	Paclitaxel is necessary for improved survival in epithelial ovarian cancers with homologous recombination gene mutations. <i>Oncotarget</i> , 2016 , 7, 48577-48585	3.3	3
192	Cancer susceptibility mutations in individuals with breast and ovarian cancer using next-generation sequencing.. <i>Journal of Clinical Oncology</i> , 2016 , 34, 1515-1515	2.2	0
191	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , 2016 , 11, e0158801	3.7	7
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30	The von Hippel-Lindau (VHL) germline mutation V84L manifests as early-onset bilateral pheochromocytoma. <i>American Journal of Medical Genetics, Part A</i> , 2006 , 140, 685-90	2.5	18
29	Childhood cancer in families with and without BRCA1 or BRCA2 mutations ascertained at a high-risk breast cancer clinic. <i>Cancer Biology and Therapy</i> , 2006 , 5, 1098-102	4.6	23
28	Genome-wide linkage screen for testicular germ cell tumour susceptibility loci. <i>Human Molecular Genetics</i> , 2006 , 15, 443-51	5.6	127
27	A multicenter study of cancer incidence in CHEK2 1100delC mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2006 , 15, 2542-5	4	44
26	The tuberous sclerosis complex. <i>New England Journal of Medicine</i> , 2006 , 355, 1345-56	59.2	1328
25	Metastatic potential of melanomas defined by specific gene expression profiles with no BRAF signature. <i>Pigment Cell & Melanoma Research</i> , 2006 , 19, 290-302		378
24	The Y deletion gr/gr and susceptibility to testicular germ cell tumor. <i>American Journal of Human Genetics</i> , 2005 , 77, 1034-43	11	172
23	Adjusting the estimated proportion of breast cancer cases associated with BRCA1 and BRCA2 mutations: public health implications. <i>Genetics in Medicine</i> , 2005 , 7, 28-33	8.1	58
22	An evaluation of BRCA1 and BRCA2 founder mutations penetrance estimates for breast cancer among Ashkenazi Jewish women. <i>Genetics in Medicine</i> , 2005 , 7, 34-9	8.1	13
21	Inherited susceptibility for pediatric cancer. <i>Cancer Journal (Sudbury, Mass)</i> , 2005 , 11, 255-67	2.2	17

20	Resolving ATM haplotypes in whites. <i>American Journal of Human Genetics</i> , 2003 , 72, 1071-3	11	1
19	Variants in CHEK2 other than 1100delC do not make a major contribution to breast cancer susceptibility. <i>American Journal of Human Genetics</i> , 2003 , 72, 1023-8	11	104
18	Pheochromocytoma: the expanding genetic differential diagnosis. <i>Journal of the National Cancer Institute</i> , 2003 , 95, 1196-204	9.7	199
17	BRCA1 and BRCA2 mutation frequency in women evaluated in a breast cancer risk evaluation clinic. <i>Journal of Clinical Oncology</i> , 2002 , 20, 994-9	2.2	61
16	Low-penetrance susceptibility to breast cancer due to CHEK2(*)1100delC in noncarriers of BRCA1 or BRCA2 mutations. <i>Nature Genetics</i> , 2002 , 31, 55-9	36.3	863
15	Cancer risk estimates for BRCA1 mutation carriers identified in a risk evaluation program. <i>Journal of the National Cancer Institute</i> , 2002 , 94, 1365-72	9.7	498
14	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. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002 , 99, 827-31	11.5	64
13	CGH-targeted linkage analysis reveals a possible BRCA1 modifier locus on chromosome 5q. <i>Human Molecular Genetics</i> , 2002 , 11, 1327-32	5.6	25
12	Absence of evidence for a familial breast cancer susceptibility gene at chromosome 8p12-p22. <i>Oncogene</i> , 2000 , 19, 4170-3	9.2	32
11	Screening for genomic rearrangements in families with breast and ovarian cancer identifies BRCA1 mutations previously missed by conformation-sensitive gel electrophoresis or sequencing. <i>American Journal of Human Genetics</i> , 2000 , 67, 841-50	11	136
10	Association of HPC2/ELAC2 genotypes and prostate cancer. <i>American Journal of Human Genetics</i> , 2000 , 67, 1014-9	11	121
9	Successful use of alternate waste nitrogen agents and hemodialysis in a patient with hyperammonemic coma after heart-lung transplantation. <i>Archives of Neurology</i> , 1999 , 56, 481-4		35
8	Multiple vascular and bowel ruptures in an adolescent male with sporadic Ehlers-Danlos syndrome type IV. <i>Pediatric and Developmental Pathology</i> , 1999 , 2, 86-93	2.2	22
7	I1307K APC variant in non-Ashkenazi Jewish women affected with breast cancer 1999 , 85, 189-190		5
6	The APC1307K allele and breast cancer risk. <i>Nature Genetics</i> , 1998 , 20, 13-4	36.3	59
5	Intestinal perforation in Ehlers-Danlos syndrome after enema treatment for constipation. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 1998 , 27, 599-602	2.8	13
4	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
3	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2

2	Assessment of Polygenic Architecture and Risk Prediction based on Common Variants Across Fourteen Cancers	1
1	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses	2