Sara Gutirrez-Enrquez

List of Publications by Citations

71 papers	1,844 citations	23 h-index	41 g-index
77 ext. papers	2,384 ext. citations	4.6 avg, IF	3.58 L-index

#	Paper	IF	Citations
71	A TP53 polymorphism is associated with increased risk of colorectal cancer and with reduced levels of TP53 mRNA. *Oncogene*, **2004**, 23, 1954-6	9.2	162
70	RAD51 foci as a functional biomarker of homologous recombination repair and PARP inhibitor resistance in germline BRCA-mutated breast cancer. *Annals of Oncology*, **2018**, 29, 1203-1210	10.3	160
69	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
68	A RAD51 assay feasible in routine tumor samples calls PARP inhibitor response beyond BRCA mutation. *EMBO Molecular Medicine*, **2018**, 10,	12	85
67	Individual patient data meta-analysis shows a significant association between the ATM rs1801516 SNP and toxicity after radiotherapy in 5456 breast and prostate cancer patients. *Radiotherapy and Oncology*, **2016**, 121, 431-439	5.3	69
66	Functional consequences of ATM sequence variants for chromosomal radiosensitivity. *Genes Chromosomes and Cancer*, **2004**, 40, 109-19	5	69
65	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium. *Human Molecular Genetics*, **2014**, 23, 3666-80	5.6	60
64	Genomic rearrangements at the BRCA1 locus in Spanish families with breast/ovarian cancer. *Clinical Chemistry*, **2006**, 52, 1480-5	5.5	54
63	Comparison of mRNA splicing assay protocols across multiple laboratories: recommendations for best practice in standardized clinical testing. *Clinical Chemistry*, **2014**, 60, 341-52	5.5	53
62	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
61	Individual patient data meta-analysis shows no association between the SNP rs1800469 in TGFB and late radiotherapy toxicity. *Radiotherapy and Oncology*, **2012**, 105, 289-95	5.3	52
60	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. *Breast Cancer Research and Treatment*, **2012**, 132, 1009-23	4.4	51
59	Cytogenetic analysis of Greek farmers using the micronucleus assay in peripheral lymphocytes and buccal cells. *Mutagenesis*, **2001**, 16, 539-45	2.8	47
58	Screening for large rearrangements of the BRCA2 gene in Spanish families with breast/ovarian cancer. *Breast Cancer Research and Treatment*, **2007**, 103, 103-7	4.4	38
57	Naturally occurring BRCA2 alternative mRNA splicing events in clinically relevant samples. *Journal of Medical Genetics*, **2016**, 53, 548-58	5.8	37
56	Spectrum of BRCA1/2 point mutations and genomic rearrangements in high-risk breast/ovarian cancer Chilean families. *Breast Cancer Research and Treatment*, **2011**, 126, 705-16	4.4	36
55	Analysis of PALB2 gene in BRCA1/BRCA2 negative Spanish hereditary breast/ovarian cancer families with pancreatic cancer cases. *PLoS ONE*, **2013**, 8, e67538	3.7	35

54	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. *Frontiers in Genetics*, **2018**, 9, 366	4.5	35
53	Multigene panel testing beyond BRCA1/2 in breast/ovarian cancer Spanish families and clinical actionability of findings. *Journal of Cancer Research and Clinical Oncology*, **2018**, 144, 2495-2513	4.9	31
52	A follow-up study on micronucleus frequency in Spanish agricultural workers exposed to pesticides. *Mutagenesis*, **2002**, 17, 79-82	2.8	29
51	REQUITE: A prospective multicentre cohort study of patients undergoing radiotherapy for breast, lung or prostate cancer. *Radiotherapy and Oncology*, **2019**, 138, 59-67	5.3	26
50	Use of the cytokinesis-block micronucleus assay to measure radiation-induced chromosome damage in lymphoblastoid cell lines. *Mutation Research - Genetic Toxicology and Environmental Mutagenesis*, **2003**, 535, 1-13	3	26
49	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. *Nature Communications*, **2018**, 9, 967	17.4	23
48	RAD51C germline mutations found in Spanish site-specific breast cancer and breast-ovarian cancer families. *Breast Cancer Research and Treatment*, **2014**, 147, 133-43	4.4	23
47	Opportunistic testing of BRCA1, BRCA2 and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. *International Journal of Cancer*, **2019**, 145, 2682-2691	7.5	21
46	About 1% of the breast and ovarian Spanish families testing negative for BRCA1 and BRCA2 are carriers of RAD51D pathogenic variants. *International Journal of Cancer*, **2014**, 134, 2088-97	7.5	21
45	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. *Breast Cancer Research and Treatment*, **2011**, 127, 671-9	4.4	21
44	A cytogenetic follow-up study of thyroid cancer patients treated with 131I. *Cancer Letters*, **1995**, 91, 199-204	3.9	21
43	Application of the single cell gel electrophoresis (SCGE) assay to the detection of DNA damage induced by 131I treatment in hyperthyroidism patients. *Mutagenesis*, **1998**, 13, 95-8	2.8	20
42	Heterogeneous prevalence of recurrent BRCA1 and BRCA2 mutations in Spain according to the geographical area: implications for genetic testing. *Familial Cancer*, **2010**, 9, 187-91	3	19
41	Ionizing radiation or mitomycin-induced micronuclei in lymphocytes of BRCA1 or BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2011**, 127, 611-22	4.4	17
40	ATM germline mutations in Spanish early-onset breast cancer patients negative for BRCA1/BRCA2 mutations. *Clinical Genetics*, **2008**, 73, 465-73	4	17
39	Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. *Clinical and Translational Oncology*, **2016**, 18, 981-7	3.6	16
38	Capillary electrophoresis analysis of conventional splicing assays: IARC analytical and clinical classification of 31 BRCA2 genetic variants. *Human Mutation*, **2014**, 35, 53-7	4.7	16
37	Screening of deep intronic regions by targeted gene sequencing identifies the first germline variant causing pseudoexon activation in a patient with breast/ovarian cancer. *Journal of Medical Genetics*, **2019**, 56, 63-74	5.8	16

36	Comparative study of chromosome aberrations induced with aphidicolin in women affected by breast cancer and cervix uterine cancer. *Cancer Genetics and Cytogenetics*, **1997**, 94, 120-4		14
35	Caution should be used when interpreting alterations affecting the exon 3 of the BRCA2 gene in breast/ovarian cancer families. *Journal of Clinical Oncology*, **2007**, 25, 5035-6; author reply 5036-8	2.2	14
34	Targeted RNA-seq successfully identifies normal and pathogenic splicing events in breast/ovarian cancer susceptibility and Lynch syndrome genes. *International Journal of Cancer*, **2019**, 145, 401-414	7.5	14
33	Assessment of blind predictions of the clinical significance of BRCA1 and BRCA2 variants. *Human Mutation*, **2019**, 40, 1546-1556	4.7	13
32	BRCA1 and BRCA2 5' noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. *Human Mutation*, **2018**, 39, 2025-2039	4.7	12
31	Characterization of four novel BRCA2 large genomic rearrangements in Spanish breast/ovarian cancer families: review of the literature, and reevaluation of the genetic mechanisms involved in their origin. *Breast Cancer Research and Treatment*, **2012**, 133, 273-83	4.4	11
30	Apoptosis for prediction of radiotherapy late toxicity: lymphocyte subset sensitivity and potential effect of TP53 Arg72Pro polymorphism. *Apoptosis: an International Journal on Programmed Cell Death*, **2015**, 20, 371-82	5.4	10
29	Mitotic delay in lymphocytes from BRCA1 heterozygotes unable to reduce the radiation-induced chromosomal damage. *DNA Repair*, **2008**, 7, 1907-11	4.3	10
28	Detection of the CHEK2 1100delC mutation by MLPA BRCA1/2 analysis: a worthwhile strategy for its clinical applicability in 1100delC low-frequency populations?. *Breast Cancer Research and Treatment*, **2008**, 107, 455-7	4.4	10
27	Novel BRCA1 deleterious mutation (c.1949_1950delTA) in a woman of Senegalese descent with triple-negative early-onset breast cancer. *Oncology Letters*, **2011**, 2, 1287-1289	2.6	8
26	The variants BRCA1 IVS6-1G>A and BRCA2 IVS15+1G>A lead to aberrant splicing of the transcripts. *Breast Cancer Research and Treatment*, **2009**, 117, 461-5	4.4	8
25	Low sensitivity of the sister chromatid exchange assay to detect the genotoxic effects of radioiodine therapy. *Mutagenesis*, **1999**, 14, 221-6	2.8	8
24	A novel de novo BRCA2 mutation of paternal origin identified in a Spanish woman with early onset bilateral breast cancer. *Breast Cancer Research and Treatment*, **2010**, 121, 221-5	4.4	7
23	Telomeric association in women with breast and uterine cervix cancer. *Cancer Genetics and Cytogenetics*, **1997**, 98, 115-8		7
22	Incorporation of semi-quantitative analysis of splicing alterations for the clinical interpretation of variants in BRCA1 and BRCA2 genes. *Human Mutation*, **2019**, 40, 2296-2317	4.7	6
21	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. *Breast Cancer Research and Treatment*, **2011**, 128, 573-9	4.4	6
20	External Validation of a Predictive Model for Acute Skin Radiation Toxicity in the REQUITE Breast Cohort. *Frontiers in Oncology*, **2020**, 10, 575909	5.3	5
19	First international workshop of the ATM and cancer risk group (4-5 December 2019). *Familial Cancer* , **2021**, 1	3	5

18	Alternative transcript imbalance underlying breast cancer susceptibility in a family carrying PALB2 c.3201+5G>T. *Breast Cancer Research and Treatment*, **2019**, 174, 543-550	4.4	5
17	Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing: BRCA2 c.7976+5G´>´T as a case study. *Human Mutation*, **2018**, 39, 1155-1160	4.7	5
16	BRCA1- and BRCA2-specific in silico tools for variant interpretation in the CAGI 5 ENIGMA challenge. *Human Mutation*, **2019**, 40, 1593-1611	4.7	4
15	Mutation analysis of the SHFM1 gene in breast/ovarian cancer families. *Journal of Cancer Research and Clinical Oncology*, **2013**, 139, 529-32	4.9	4
14	Preclinical In Vivo Validation of the RAD51 Test for Identification of Homologous Recombination-Deficient Tumors and Patient Stratification.. *Cancer Research*, **2022**, 82, 1646-1657	10.1	4
13	Mutation analysis of the BCCIP gene for breast cancer susceptibility in breast/ovarian cancer families. *Gynecologic Oncology*, **2013**, 131, 460-3	4.9	3
12	Identification of a new complex deleterious mutation in exon 18 of the BRCA2 gene in a hereditary male/female breast cancer family. *Breast Cancer Research and Treatment*, **2010**, 123, 587-90	4.4	3
11	Role of Splicing Regulatory Elements and In Silico Tools Usage in the Identification of Deep Intronic Splicing Variants in Hereditary Breast/Ovarian Cancer Genes. *Cancers*, **2021**, 13,	6.6	3
10	Evaluation of the association of heterozygous germline variants in NTHL1 with breast cancer predisposition: an international multi-center study of 47,180 subjects. *Npj Breast Cancer*, **2021**, 7, 52	7.8	2
9	External Validation of a Predictive Model for Acute Skin Radiation Toxicity in the REQUITE Breast Cohort. *Frontiers in Oncology*, **2020**, 10, 575909	5.3	1
8	A Collaborative Effort to Define Classification Criteria for ATM Variants in Hereditary Cancer Patients. *Clinical Chemistry*, **2021**, 67, 518-533	5.5	1
7	BRCA1 and BRCA2 whole cDNA analysis in unsolved hereditary breast/ovarian cancer patients. *Cancer Genetics*, **2021**, 258-259, 10-17	2.3	1
6	Development and Optimization of a Machine-Learning Prediction Model for Acute Desquamation After Breast Radiation Therapy in the Multicenter REQUITE Cohort. *Advances in Radiation Oncology*, **2022**, 7, 100890	3.3	0
5	The computational approach to variant interpretation **2021**, 89-119		0
4	Clinical consequences of BRCA2 hypomorphism. *Npj Breast Cancer*, **2021**, 7, 117	7.8	0
3	Overview of health-related quality of life and toxicity of non-small cell lung cancer patients receiving curative-intent radiotherapy in a real-life setting (the REQUITE study).. *Lung Cancer*, **2022**, 166, 228-241	5.9	0
2	Re: correspondence from Dr. Michael Swift, Disease Insight Research Foundation, concerning Gutiérrez-Enríquez S, Fernet M, Dörk T, Bremer M, Lauge A, Stoppa-Lyonnet D, Moullan N, Angèle S, Hall J, Functional consequences of the ATM sequence variants for chromosomal radiosensitivity. *Genes Chromosomes and Cancer*, **2005**, 42, 202-203	5	
1	A data science approach for early-stage prediction of Patient's susceptibility to acute side effects of advanced radiotherapy. *Computers in Biology and Medicine*, **2021**, 135, 104624	7	