Miguel de la Hoya

List of Publications by Citations

68 papers	3,027 citations	28 h-index	54 g-index
76 ext. papers	4,144 ext. citations	9.4 avg, IF	3.03 L-index

#	Paper	IF	Citations
68	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
67	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
66	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
65	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
64	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
63	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
62	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
61	Analysis of BRCA1 and BRCA2 genes in Spanish breast/ovarian cancer patients: a high proportion of mutations unique to Spain and evidence of founder effects. *Human Mutation*, **2003**, 22, 301-12	4.7	133
60	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
59	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
58	The average cumulative risks of breast and ovarian cancer for carriers of mutations in BRCA1 and BRCA2 attending genetic counseling units in Spain. *Clinical Cancer Research*, **2008**, 14, 2861-9	12.9	77
57	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65
56	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
55	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
54	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
53	Combined genetic and splicing analysis of BRCA1 c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. *Human Molecular Genetics*, **2016**, 25, 2256-2268	5.6	55
52	Genomic rearrangements at the BRCA1 locus in Spanish families with breast/ovarian cancer. *Clinical Chemistry*, **2006**, 52, 1480-5	5.5	54

51	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
50	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
49	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
48	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
47	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
46	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. *Breast Cancer Research and Treatment*, **2012**, 132, 307-15	4.4	44
45	Naturally occurring BRCA2 alternative mRNA splicing events in clinically relevant samples. *Journal of Medical Genetics*, **2016**, 53, 548-58	5.8	37
44	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
43	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
42	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
41	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
40	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
39	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
38	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
37	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
36	Association between BRCA1 mutations and ratio of female to male births in offspring of families with breast cancer, ovarian cancer, or both. *JAMA - Journal of the American Medical Association*, **2003**, 290, 929-31	27.4	24
35	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
34	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

33	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. *Journal of Medical Genetics*, **2019**, 56, 347-357	5.8	19
32	Alternative splicing and molecular characterization of splice site variants: BRCA1 c.591C>T as a case study. *Clinical Chemistry*, **2010**, 56, 53-61	5.5	19
31	The BRCA2 c.68-7T□□A variant is not pathogenic: A model for clinical calibration of spliceogenicity. *Human Mutation*, **2018**, 39, 729-741	4.7	16
30	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
29	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. *Gynecologic Oncology*, **2016**, 141, 386-401	4.9	15
28	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
27	Novel genetic mutations detected by multigene panel are associated with hereditary colorectal cancer predisposition. *PLoS ONE*, **2018**, 13, e0203885	3.7	15
26	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
25	BRCA1 Alternative splicing landscape in breast tissue samples. *BMC Cancer*, **2015**, 15, 219	4.8	13
24	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
23	Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report. *Journal of Medical Genetics*, **2019**, 56, 453-460	5.8	10
22	RECQL5: Another DNA helicase potentially involved in hereditary breast cancer susceptibility. *Human Mutation*, **2019**, 40, 566-577	4.7	10
21	Association of a let-7 miRNA binding region of TGFBR1 with hereditary mismatch repair proficient colorectal cancer (MSS HNPCC). *Carcinogenesis*, **2016**, 37, 751-8	4.6	9
20	Role of GALNT12 in the genetic predisposition to attenuated adenomatous polyposis syndrome. *PLoS ONE*, **2017**, 12, e0187312	3.7	8
19	Comprehensive Assessment of Messenger Ribonucleic Acid Splicing With Implications for Variant Classification. *Frontiers in Genetics*, **2019**, 10, 1139	4.5	8
18	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. *Familial Cancer*, **2015**, 14, 505-13	3	7
17	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
16	Alternative mRNA splicing can attenuate the pathogenicity of presumed loss-of-function variants in BRCA2. *Genetics in Medicine*, **2020**, 22, 1355-1365	8.1	6

List of Publications

15	Thorough in silico and in vitro cDNA analysis of 21 putative BRCA1 and BRCA2 splice variants and a complex tandem duplication in BRCA2 allowing the identification of activated cryptic splice donor sites in BRCA2 exon 11. *Human Mutation*, **2018**, 39, 515-526	4.7	5
14	Contribution of New Adenomatous Polyposis Predisposition Genes in an Unexplained Attenuated Spanish Cohort by Multigene Panel Testing. *Scientific Reports*, **2019**, 9, 9814	4.9	5
13	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
12	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. *British Journal of Cancer*, **2017**, 117, 1048-1062	8.7	4
11	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
10	Mutant BRCA1 alleles transmission: different approaches and different biases. *International Journal of Cancer*, **2005**, 113, 166-7	7.5	2
9	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants.. *Genetics in Medicine*, **2021**,	8.1	2
8	Response. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	2
7	Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the Gene. *Cancers*, **2020**, 12,	6.6	1
6	Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. *Breast Cancer Research*, **2020**, 22, 108	8.3	0
5	, a Gene Potentially Implicated in Familial Colorectal Cancer Type X. *Cancer Prevention Research*, **2021**, 14, 185-194	3.2	0
4	Altered regulation of BRCA1 exon 11 splicing is associated with breast cancer risk in carriers of BRCA1 pathogenic variants. *Human Mutation*, **2021**, 42, 1488-1502	4.7	0
3	Breast cancer risks associated with missense variants in breast cancer susceptibility genes.. *Genome Medicine*, **2022**, 14, 51	14.4	0
2	Lack of germ-line mutations at the specific BRCA1-IRIS coding sequence in 114 Spanish high-risk breast/ovarian families. *Familial Cancer*, **2005**, 4, 317-9	3	
1	Functional evidence (I) transcripts and RNA-splicing outline **2021**, 121-144		