

Miguel de la Hoya

List of Publications by Year in descending order

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73
papers

4,848
citations

136740

32
h-index

102304

66
g-index

76
all docs

76
docs citations

76
times ranked

7414
citing authors

#	ARTICLE	IF	CITATIONS
1	Breast Cancer Risk Genes " Association Analysis in More than 113,000 Women. <i>New England Journal of Medicine</i> , 2021, 384, 428-439.	13.9	532
2	Association of Type and Location of <i>BRCA1</i> and <i>BRCA2</i> Mutations With Risk of Breast and Ovarian Cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015, 313, 1347.	3.8	390
3	A locus on 19p13 modifies risk of breast cancer in <i>BRCA1</i> mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. <i>Nature Genetics</i> , 2010, 42, 885-892.	9.4	309
4	Genome-Wide Association Study in <i>BRCA1</i> Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003212.	1.5	244
5	Mutational spectrum in a worldwide study of 29,700 families with <i>BRCA1</i> or <i>BRCA2</i> mutations. <i>Human Mutation</i> , 2018, 39, 593-620.	1.1	224
6	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015, 47, 164-171.	9.4	221
7	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Implications for Risk Prediction. <i>Cancer Research</i> , 2010, 70, 9742-9754.	0.4	169
8	Analysis of <i>BRCA1</i> and <i>BRCA2</i> genes in Spanish breast/ovarian cancer patients: A high proportion of mutations unique to Spain and evidence of founder effects. <i>Human Mutation</i> , 2003, 22, 301-312.	1.1	154
9	Prediction of Breast and Prostate Cancer Risks in Male <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017, 35, 2240-2250.	0.8	152
10	Breast cancer risk variants at 6q25 display different phenotype associations and regulate <i>ESR1</i> , <i>RMND1</i> and <i>CCDC170</i> . <i>Nature Genetics</i> , 2016, 48, 374-386.	9.4	125
11	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
12	Combined genetic and splicing analysis of <i>BRCA1</i> c.[594-2A>C; 641A>G] highlights the relevance of naturally occurring in-frame transcripts for developing disease gene variant classification algorithms. <i>Human Molecular Genetics</i> , 2016, 25, 2256-2268.	1.4	106
13	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in <i>RAD51C</i> and <i>RAD51D</i> . <i>Journal of the National Cancer Institute</i> , 2020, 112, 1242-1250.	3.0	106
14	Large scale multifactorial likelihood quantitative analysis of <i>BRCA1</i> and <i>BRCA2</i> variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019, 40, 1557-1578.	1.1	102
15	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the <i>BRCA1</i> locus: a report from the ENIGMA consortium. <i>Human Molecular Genetics</i> , 2014, 23, 3666-3680.	1.4	96
16	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. <i>Clinical Chemistry</i> , 2014, 60, 341-352.	1.5	95
17	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	5.8	93
18	The Average Cumulative Risks of Breast and Ovarian Cancer for Carriers of Mutations in <i>BRCA1</i> and <i>BRCA2</i> Attending Genetic Counseling Units in Spain. <i>Clinical Cancer Research</i> , 2008, 14, 2861-2869.	3.2	90

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19	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019, 10, 1741.	5.8	90
20	Cancer Risks Associated With <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>Journal of Clinical Oncology</i> , 2022, 40, 1529-1541.	0.8	90
21	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of <i>BRCA1</i> and <i>BRCA2</i> pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1653-1666.	1.1	82
22	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016, 7, 12675.	5.8	78
23	<i>BRCA2</i> Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016, 108, djv315.	3.0	77
24	Naturally occurring <i>BRCA2</i> alternative mRNA splicing events in clinically relevant samples. <i>Journal of Medical Genetics</i> , 2016, 53, 548-558.	1.5	69
25	Genomic Rearrangements at the <i>BRCA1</i> Locus in Spanish Families with Breast/Ovarian Cancer. <i>Clinical Chemistry</i> , 2006, 52, 1480-1485.	1.5	60
26	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. <i>Frontiers in Genetics</i> , 2018, 9, 366.	1.1	53
27	Detection of a large rearrangement in <i>PALB2</i> in Spanish breast cancer families with male breast cancer. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 307-315.	1.1	50
28	Characterization of the Cancer Spectrum in Men With Germline <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2020, 6, 1218.	3.4	48
29	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>PLoS Genetics</i> , 2014, 10, e1004256.	1.5	47
30	Analysis of <i>PALB2</i> Gene in <i>BRCA1/BRCA2</i> Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. <i>PLoS ONE</i> , 2013, 8, e67538.	1.1	44
31	Inheritance of deleterious mutations at both <i>BRCA1</i> and <i>BRCA2</i> in an international sample of 32,295 women. <i>Breast Cancer Research</i> , 2016, 18, 112.	2.2	42
32	Association of Genomic Domains in <i>BRCA1</i> and <i>BRCA2</i> with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020, 80, 624-638.	0.4	39
33	Assessing Associations between the <i>AURKA-HMMR-TPX2-TUBG1</i> Functional Module and Breast Cancer Risk in <i>BRCA1/2</i> Mutation Carriers. <i>PLoS ONE</i> , 2015, 10, e0120020.	1.1	34
34	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. <i>Journal of Medical Genetics</i> , 2019, 56, 347-357.	1.5	32
35	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. <i>Breast Cancer Research</i> , 2016, 18, 64.	2.2	31
36	Association Between <i>BRCA1</i> Mutations and Ratio of Female to Male Births in Offspring of Families With Breast Cancer, Ovarian Cancer, or Both. <i>JAMA - Journal of the American Medical Association</i> , 2003, 290, 929.	3.8	30

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37	Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report. <i>Journal of Medical Genetics</i> , 2019, 56, 453-460.	1.5	30
38	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	2.3	28
39	Targeted RNA-seq successfully identifies normal and pathogenic splicing events in breast/ovarian cancer susceptibility and Lynch syndrome genes. <i>International Journal of Cancer</i> , 2019, 145, 401-414.	2.3	27
40	Capillary Electrophoresis Analysis of Conventional Splicing Assays: IARC Analytical and Clinical Classification of 31 BRCA2 Genetic Variants. <i>Human Mutation</i> , 2014, 35, 53-57.	1.1	25
41	Novel genetic mutations detected by multigene panel are associated with hereditary colorectal cancer predisposition. <i>PLoS ONE</i> , 2018, 13, e0203885.	1.1	24
42	Alternative mRNA splicing can attenuate the pathogenicity of presumed loss-of-function variants in BRCA2. <i>Genetics in Medicine</i> , 2020, 22, 1355-1365.	1.1	23
43	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. <i>European Journal of Human Genetics</i> , 2022, 30, 349-362.	1.4	23
44	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2015, 24, 308-316.	1.1	22
45	Alternative Splicing and Molecular Characterization of Splice Site Variants: BRCA1 c.591C>T as a Case Study. <i>Clinical Chemistry</i> , 2010, 56, 53-61.	1.5	21
46	The BRCA2 c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018, 39, 729-741.	1.1	19
47	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> , 2022, 114, 109-122.	3.0	19
48	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. <i>Genome Medicine</i> , 2022, 14, 51.	3.6	19
49	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016, 141, 386-401.	0.6	18
50	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.	1.1	18
51	BRCA1 Alternative splicing landscape in breast tissue samples. <i>BMC Cancer</i> , 2015, 15, 219.	1.1	17
52	Association of a let-7 miRNA binding region of TGFBR1 with hereditary mismatch repair proficient colorectal cancer (MSS HNPCC). <i>Carcinogenesis</i> , 2016, 37, 751-758.	1.3	16
53	RECQL5 : Another DNA helicase potentially involved in hereditary breast cancer susceptibility. <i>Human Mutation</i> , 2019, 40, 566-577.	1.1	16
54	Splicing predictions, minigene analyses, and ACMG-AMP clinical classification of 42 germline PALB2 splice site variants. <i>Journal of Pathology</i> , 2022, 256, 321-334.	2.1	16

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55	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. <i>Familial Cancer</i> , 2015, 14, 505-513.	0.9	15
56	Characterisation of the novel deleterious RAD51C p.Arg312Trp variant and prioritisation criteria for functional analysis of RAD51C missense changes. <i>British Journal of Cancer</i> , 2017, 117, 1048-1062.	2.9	12
57	Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing: <i>BRCA2</i> c.7976+5G>A as a case study. <i>Human Mutation</i> , 2018, 39, 1155-1160.	1.1	12
58	Role of GALNT12 in the genetic predisposition to attenuated adenomatous polyposis syndrome. <i>PLoS ONE</i> , 2017, 12, e0187312.	1.1	10
59	Comprehensive Assessment of BARD1 Messenger Ribonucleic Acid Splicing With Implications for Variant Classification. <i>Frontiers in Genetics</i> , 2019, 10, 1139.	1.1	10
60	Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the RAD51C Gene. <i>Cancers</i> , 2020, 12, 3771.	1.7	10
61	RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. <i>Cancers</i> , 2021, 13, 2845.	1.7	10
62	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants. <i>Genetics in Medicine</i> , 2022, 24, 119-129.	1.1	10
63	Contribution of New Adenomatous Polyposis Predisposition Genes in an Unexplained Attenuated Spanish Cohort by Multigene Panel Testing. <i>Scientific Reports</i> , 2019, 9, 9814.	1.6	9
64	Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. <i>Breast Cancer Research</i> , 2020, 22, 108.	2.2	9
65	<i>BRIP1</i> , a Gene Potentially Implicated in Familial Colorectal Cancer Type X. <i>Cancer Prevention Research</i> , 2021, 14, 185-194.	0.7	7
66	Altered regulation of <i>BRCA1</i> exon 11 splicing is associated with breast cancer risk in carriers of <i>BRCA1</i> pathogenic variants. <i>Human Mutation</i> , 2021, 42, 1488-1502.	1.1	7
67	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. <i>Human Mutation</i> , 2018, 39, 515-526.	1.1	5
68	Minigene-based splicing analysis and ACMG/AMP-based tentative classification of 56 <i>ATM</i> variants. <i>Journal of Pathology</i> , 2022, 258, 83-101.	2.1	5
69	Mutant BRCA1 alleles transmission: Different approaches and different biases. <i>International Journal of Cancer</i> , 2005, 113, 166-167.	2.3	4
70	Minigene Splicing Assays Identify 20 Spliceogenic Variants of the Breast/Ovarian Cancer Susceptibility Gene RAD51C. <i>Cancers</i> , 2022, 14, 2960.	1.7	3
71	Response: Table 1.. <i>Journal of the National Cancer Institute</i> , 2016, 108, djw173.	3.0	2
72	Lack of Germ-line Mutations at the Specific BRCA1-IRIS Coding Sequence in 114 Spanish High-risk Breast/ovarian Families. <i>Familial Cancer</i> , 2005, 4, 317-319.	0.9	0

#	ARTICLE	IF	CITATIONS
73	Functional evidence (l)Âtranscripts and RNA-splicing outline. , 2021, , 121-144.		0