Miguel de la Hoya

List of Publications by Year
in descending order

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73 papers	4,848 citations	136950 32 h-index	102487 66 g-index
76 all docs	76 docs citations	76 times ranked	7414 citing authors

#	Article	IF	Citations
1	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
2	Splicing predictions, minigene analyses, and <scp>ACMG</scp>â€<scp>AMP</scp> clinical classification of 42 germline <scp>PALB2</scp> spliceâ€site variants. Journal of Pathology, 2022, 256, 321-334.	4.5	16
3	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, 2022, 24, 119-129.	2.4	10
4	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
5	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
6	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	8.2	19
7	Minigeneâ€based splicing analysis and <scp>ACMG</scp>/<scp>AMP</scp>â€based tentative classification of 56 <scp>ATM</scp> variants. Journal of Pathology, 2022, 258, 83-101.	4.5	5
8	Minigene Splicing Assays Identify 20 Spliceogenic Variants of the Breast/Ovarian Cancer Susceptibility Gene RAD51C. Cancers, 2022, 14, 2960.	3.7	3
9	BRIP1, a Gene Potentially Implicated in Familial Colorectal Cancer Type X. Cancer Prevention Research, 2021, 14, 185-194.	1.5	7
10	Functional evidence (I)Âtranscripts and RNA-splicing outline. , 2021, , 121-144.		0
11	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	27.0	532
12	RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. Cancers, 2021, 13, 2845.	3.7	10
13	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.	2.5	7
14	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
15	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
16	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.	5.0	9
17	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.	2.4	82
18	Comprehensive Functional Characterization and Clinical Interpretation of 20 Splice-Site Variants of the RAD51C Gene. Cancers, 2020, 12, 3771.	3.7	10

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19	Alternative mRNA splicing can attenuate the pathogenicity of presumed loss-of-function variants in BRCA2. Genetics in Medicine, 2020, 22, 1355-1365.	2.4	23
20	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
21	Ovarian and Breast Cancer Risks Associated With Pathogenic Variants in RAD51C and RAD51D. Journal of the National Cancer Institute, 2020, 112, 1242-1250.	6.3	106
22	Contribution of New Adenomatous Polyposis Predisposition Genes in an Unexplained Attenuated Spanish Cohort by Multigene Panel Testing. Scientific Reports, 2019, 9, 9814.	3.3	9
23	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
24	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.	2.5	102
25	Towards controlled terminology for reporting germline cancer susceptibility variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 347-357.	3.2	32
26	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
27	Alternative splicing and ACMG-AMP-2015-based classification of PALB2 genetic variants: an ENIGMA report. Journal of Medical Genetics, 2019, 56, 453-460.	3.2	30
28	RECQL5 : Another DNA helicase potentially involved in hereditary breast cancer susceptibility. Human Mutation, 2019, 40, 566-577.	2.5	16
29	Comprehensive Assessment of BARD1 Messenger Ribonucleic Acid Splicing With Implications for Variant Classification. Frontiers in Genetics, 2019, 10, 1139.	2.3	10
30	Targeted RNAâ€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.	5.1	27
31	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	2.5	19
32	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
33	Thorough in silico and in vitro cDNA analysis of 21 putativeBRCA1andBRCA2splice variants and a complex tandem duplication inBRCA2allowing the identification of activated cryptic splice donor sites inBRCA2exon 11. Human Mutation, 2018, 39, 515-526.	2.5	5
34	Novel genetic mutations detected by multigene panel are associated with hereditary colorectal cancer predisposition. PLoS ONE, 2018, 13, e0203885.	2.5	24
35	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.	2.3	53
36	Characterization of spliceogenic variants located in regions linked to high levels of alternative splicing:BRCA2c.7976+5GÂ>ÂT as a case study. Human Mutation, 2018, 39, 1155-1160.	2.5	12

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37	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.	6.4	12
38	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.	2.5	18
39	Role of GALNT12 in the genetic predisposition to attenuated adenomatous polyposis syndrome. PLoS ONE, 2017, 12, e0187312.	2.5	10
40	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.	1.6	152
41	Naturally occurringBRCA2alternative mRNA splicing events in clinically relevant samples. Journal of Medical Genetics, 2016, 53, 548-558.	3.2	69
42	Response: Table 1.. Journal of the National Cancer Institute, 2016, 108, djw173.	6.3	2
43	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.	5.0	31
44	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	5.0	42
45	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
46	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
47	Association of a let-7 miRNA binding region ofTGFBR1with hereditary mismatch repair proficient colorectal cancer (MSS HNPCC). Carcinogenesis, 2016, 37, 751-758.	2.8	16
48	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.	2.9	106
49	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. Nature Genetics, 2016, 48, 374-386.	21.4	125
50	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
51	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18
52	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
53	BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study. Familial Cancer, 2015, 14, 505-513.	1.9	15
54	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221

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55	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
56	BRCA1 Alternative splicing landscape in breast tissue samples. BMC Cancer, 2015, 15, 219.	2.6	17
57	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22
58	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
59	Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium. Human Molecular Genetics, 2014, 23, 3666-3680.	2.9	96
60	Capillary Electrophoresis Analysis of Conventional Splicing Assays: IARC Analytical and Clinical Classification of 31BRCA2Genetic Variants. Human Mutation, 2014, 35, 53-57.	2.5	25
61	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	3.2	95
62	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
63	Analysis of PALB2 Gene in BRCA1/BRCA2 Negative Spanish Hereditary Breast/Ovarian Cancer Families with Pancreatic Cancer Cases. PLoS ONE, 2013, 8, e67538.	2.5	44
64	Detection of a large rearrangement in PALB2 in Spanish breast cancer families with male breast cancer. Breast Cancer Research and Treatment, 2012, 132, 307-315.	2.5	50
65	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	21.4	309
66	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, 9742-9754.	0.9	169
67	Alternative Splicing and Molecular Characterization of Splice Site Variants: BRCA1 c.591C>T as a Case Study. Clinical Chemistry, 2010, 56, 53-61.	3.2	21
68	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, 2861-2869.	7.0	90
69	Genomic Rearrangements at the BRCA1 Locus in Spanish Families with Breast/Ovarian Cancer. Clinical Chemistry, 2006, 52, 1480-1485.	3.2	60
70	Mutant BRCA1 alleles transmission: Different approaches and different biases. International Journal of Cancer, 2005, 113, 166-167.	5.1	4
71	Lack of Germ-line Mutations at the Specific BRCA1-IRIS Coding Sequence in 114 Spanish High-risk Breast/ovarian Families. Familial Cancer, 2005, 4, 317-319.	1.9	0
72	Analysis ofBRCA1andBRCA2genes in Spanish breast/ovarian cancer patients: A high proportion of mutations unique to Spain and evidence of founder effects. Human Mutation, 2003, 22, 301-312.	2.5	154

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73	Association Between <EMPH TYPE="ITAL">BRCA1</EMPH> 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.	7.4	30