Marco Montagna

List of Publications by Citations

121 papers	7,049 citations	46 h-index	82 g-index
126 ext. papers	8,474 ext. citations	8.9 avg, IF	4.06 L-index

#	Paper	IF	Citations
121	Association between BRCA1 and BRCA2 mutations and survival in women with invasive epithelial ovarian cancer. *JAMA - Journal of the American Medical Association*, **2012**, 307, 382-90	27.4	427
120	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
119	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 134-47	4	411
118	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
117	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
116	ENIGMA--evidence-based network for the interpretation of germline mutant alleles: an international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. *Human Mutation*, **2012**, 33, 2-7	4.7	211
115	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
114	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
113	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
112	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
111	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. *Journal of the National Cancer Institute*, **2017**, 109,	9.7	153
110	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
109	Genomic rearrangements account for more than one-third of the BRCA1 mutations in northern Italian breast/ovarian cancer families. *Human Molecular Genetics*, **2003**, 12, 1055-61	5.6	144
108	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
107	Association between MDM2-SNP309 and age at colorectal cancer diagnosis according to p53 mutation status. *Journal of the National Cancer Institute*, **2006**, 98, 285-8	9.7	118
106	Germline mutation in BRCA1 or BRCA2 and ten-year survival for women diagnosed with epithelial ovarian cancer. *Clinical Cancer Research*, **2015**, 21, 652-7	12.9	107
105	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

104	The human plakoglobin gene localizes on chromosome 17q21 and is subjected to loss of heterozygosity in breast and ovarian cancers. *Proceedings of the National Academy of Sciences of the United States of America*, **1995**, 92, 6384-8	11.5	98
103	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
102	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
101	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. *American Journal of Human Genetics*, **2000**, 67, 207-212	11	89
100	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. *Nature Communications*, **2013**, 4, 1627	17.4	85
99	Identification of a 3 kb Alu-mediated BRCA1 gene rearrangement in two breast/ovarian cancer families. *Oncogene*, **1999**, 18, 4160-5	9.2	85
98	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
97	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
96	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
95	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
94	FANCM c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. *Human Molecular Genetics*, **2015**, 24, 5345-55	5.6	68
93	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
92	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
91	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2011**, 13, R110	8.3	62
90	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 3304-21	5.6	62
89	Clinical and pathologic characteristics of BRCA-positive and BRCA-negative male breast cancer patients: results from a collaborative multicenter study in Italy. *Breast Cancer Research and Treatment*, **2012**, 134, 411-8	4.4	58
88	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2016**, 18, 15	8.3	58
87	Altered tumor formation and evolutionary selection of genetic variants in the human MDM4 oncogene. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 10236-41	11.5	56

86	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
85	Color bar coding the BRCA1 gene on combed DNA: a useful strategy for detecting large gene rearrangements. *Genes Chromosomes and Cancer*, **2001**, 31, 75-84	5	55
84	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
83	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
82	Role of beta-defensin-1 polymorphisms in mother-to-child transmission of HIV-1. *Journal of Acquired Immune Deficiency Syndromes (1999)*, **2009**, 51, 13-9	3.1	53
81	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
80	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
79	Large genomic deletions inactivate the BRCA2 gene in breast cancer families. *Journal of Medical Genetics*, **2005**, 42, e64	5.8	51
78	Evaluation of widely used models for predicting BRCA1 and BRCA2 mutations. *Journal of Medical Genetics*, **2004**, 41, 278-85	5.8	50
77	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
76	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2014**, 16, 3416	8.3	46
75	Penetrances of breast and ovarian cancer in a large series of families tested for BRCA1/2 mutations. *European Journal of Human Genetics*, **2004**, 12, 899-906	5.3	46
74	Lack of association between androgen receptor CAG polymorphism and familial breast/ovarian cancer. *Cancer Letters*, **2001**, 168, 31-6	9.9	46
73	Shared heritability and functional enrichment across six solid cancers. *Nature Communications*, **2019** , 10, 431	17.4	45
72	The role of KRAS rs61764370 in invasive epithelial ovarian cancer: implications for clinical testing. *Clinical Cancer Research*, **2011**, 17, 3742-50	12.9	45
71	Prevalence of BRCA1 genomic rearrangements in a large cohort of Italian breast and breast/ovarian cancer families without detectable BRCA1 and BRCA2 point mutations. *Genes Chromosomes and Cancer*, **2006**, 45, 791-7	5	45
70	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 645-57	4	44
69	Epstein-Barr virus-associated post-transplant lympho-proliferative disease of donor origin in liver transplant recipients. *Journal of Hepatology*, **1997**, 26, 926-34	13.4	41

68	BRCA1 p.Val1688del is a deleterious mutation that recurs in breast and ovarian cancer families from Northeast Italy. *Journal of Clinical Oncology*, **2008**, 26, 26-31	2.2	39
67	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
66	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. *Human Mutation*, **2004**, 24, 100-1	4.7	36
65	Toll-like receptor 9 polymorphisms influence mother-to-child transmission of human immunodeficiency virus type 1. *Journal of Translational Medicine*, **2010**, 8, 49	8.5	35
64	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
63	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
62	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. *Cancer Research*, **2018**, 78, 5419-5430	10.1	32
61	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
60	Association of low-penetrance alleles with male breast cancer risk and clinicopathological characteristics: results from a multicenter study in Italy. *Breast Cancer Research and Treatment*, **2013**, 138, 861-8	4.4	26
59	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
58	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
57	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
56	Occasional loss of constitutive heterozygosity at 11p15.5 and imprinting relaxation of the IGFII maternal allele in hepatoblastoma. *Journal of Cancer Research and Clinical Oncology*, **1994**, 120, 732-6	4.9	25
55	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
54	Establishment and characterization of xenografts and cancer cell cultures derived from BRCA1 -/- epithelial ovarian cancers. *European Journal of Cancer*, **2006**, 42, 1475-83	7.5	24
53	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
52	Whole-exome sequencing and targeted gene sequencing provide insights into the role of PALB2 as a male breast cancer susceptibility gene. *Cancer*, **2017**, 123, 210-218	6.4	22
51	DNA copy number alterations correlate with survival of esophageal adenocarcinoma patients. *Modern Pathology*, **2009**, 22, 58-65	9.8	22

50	Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian BRCA1/2 carriers. *Breast Cancer Research and Treatment*, **2007**, 103, 29-36	4.4	22
49	Different expressivity of BRCA1 and BRCA2: analysis of 179 Italian pedigrees with identified mutation. *Breast Cancer Research and Treatment*, **2003**, 81, 71-9	4.4	22
48	Insight into genetic susceptibility to male breast cancer by multigene panel testing: Results from a multicenter study in Italy. *International Journal of Cancer*, **2019**, 145, 390-400	7.5	22
47	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. *Journal of the National Cancer Institute*, **2019**, 111, 350-364	9.7	22
46	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
45	Novel and known genetic variants for male breast cancer risk at 8q24.21, 9p21.3, 11q13.3 and 14q24.1: results from a multicenter study in Italy. *European Journal of Cancer*, **2015**, 51, 2289-95	7.5	20
44	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
43	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1362-70	4	20
42	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
41	Performance of BOADICEA and BRCAPRO genetic models and of empirical criteria based on cancer family history for predicting BRCA mutation carrier probabilities: a retrospective study in a sample of Italian cancer genetics clinics. *Breast*, **2013**, 22, 1130-5	3.6	16
40	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. *Breast Cancer Research*, **2015**, 17, 61	8.3	16
39	Anomalous transcripts and allelic deletions of the FHIT gene in human esophageal cancer. *Cancer Genetics and Cytogenetics*, **2000**, 119, 56-61		16
38	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. *Gynecologic Oncology*, **2016**, 141, 386-401	4.9	15
37	Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for BRCA1 pathogenic variant carriers. *European Journal of Human Genetics*, **2017**, 25, 432-438	5.3	15
36	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
35	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
34	DNA copy number profile discriminates between esophageal adenocarcinoma and squamous cell carcinoma and represents an independent prognostic parameter in esophageal adenocarcinoma. *Cancer Letters*, **2011**, 310, 84-93	9.9	14
33	Contribution of Variants to Male Breast Cancer Risk: Results From a Multicenter Study in Italy. *Frontiers in Oncology*, **2018**, 8, 583	5.3	14

32	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2019**, 121, 180-192	8.7	13
31	A possible role of FANCM mutations in male breast cancer susceptibility: Results from a multicenter study in Italy. *Breast*, **2018**, 38, 92-97	3.6	13
30	CDKN2A unclassified variants in familial malignant melanoma: combining functional and computational approaches for their assessment. *Human Mutation*, **2014**, 35, 828-40	4.7	13
29	Functional impairment of p16(INK4A) due to CDKN2A p.Gly23Asp missense mutation. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2009**, 671, 26-32	3.3	13
28	Evaluation of a candidate breast cancer associated SNP in ERCC4 as a risk modifier in BRCA1 and BRCA2 mutation carriers. Results from the Consortium of Investigators of Modifiers of BRCA1/BRCA2 (CIMBA). *British Journal of Cancer*, **2009**, 101, 2048-54	8.7	13
27	Co-existence of cutaneous T-cell lymphoma and B hairy cell leukemia. *American Journal of Hematology*, **2000**, 64, 197-202	7.1	13
26	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
25	The role of genetic variants of Stromal cell-Derived Factor 1 in pediatric HIV-1 infection and disease progression. *PLoS ONE*, **2012**, 7, e44460	3.7	11
24	Contribution of susceptibility gene variants to melanoma risk in families from the Veneto region of Italy. *Pigment Cell and Melanoma Research*, **2011**, 24, 728-30	4.5	11
23	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
22	Identification of BRCA1 and BRCA2 carriers by allele-specific gene expression (AGE) analysis. *International Journal of Cancer*, **2002**, 98, 732-6	7.5	9
21	Dominance of a single Epstein-Barr virus strain in SCID-mouse tumors induced by injection of peripheral blood mononuclear cells from healthy human donors. *Virus Research*, **1995**, 36, 215-31	6.4	8
20	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
19	Family history of cancer rather than p53 status predicts efficacy of pegylated liposomal doxorubicin and oxaliplatin in relapsed ovarian cancer. *International Journal of Gynecological Cancer*, **2009**, 19, 1022-8	3.5	6
18	Analysis of Epstein-Barr virus (EBV) type and variant in spontaneous lymphoblastoid cells and Hu-SCID mouse tumours. *Molecular and Cellular Probes*, **1996**, 10, 453-61	3.3	6
17	Localization of the human phosphotyrosine phosphatase-related genes (h-PRL-1) to chromosome bands 1p35-p34, 17q12-q21, 11q24-q25 and 12q24. *Human Genetics*, **1996**, 98, 738-40	6.3	6
16	The BRCA2 sequence variant IVS19+1G-->A leads to an aberrant transcript lacking exon 19. *Cancer Genetics and Cytogenetics*, **2003**, 141, 175-6		5
15	Association of SULT1A1 Arg□□His polymorphism with male breast cancer risk: results from a multicenter study in Italy. *Breast Cancer Research and Treatment*, **2014**, 148, 623-8	4.4	4

14	Differential sensitivity of BRCA1-mutated HCC1937 human breast cancer cells to microtubule-interfering agents **2005**, 26, 1257		4
13	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
12	Evaluation of chromosome 6p22 as a breast cancer risk modifier locus in a follow-up study of BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2012**, 136, 295-302	4.4	3
11	Segregation analysis of the BRCA2 c.9227G>T variant in multiple families suggests a pathogenic role in breast and ovarian cancer predisposition. *Scientific Reports*, **2020**, 10, 13987	4.9	3
10	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
9	TP53 gene mutations in gastric carcinoma detected by polymerase chain reaction/single-strand conformation polymorphism analysis of archival material. *Journal of Cancer Research and Clinical Oncology*, **1995**, 121, 79-83	4.9	2
8	Identification of a human endogenous LTR-like sequence using HIV-1 LTR specific primers. *Molecular and Cellular Probes*, **1996**, 10, 443-51	3.3	2
7	Evaluation of CYP17A1 and CYP1B1 polymorphisms in male breast cancer risk. *Endocrine Connections*, **2019**, 8, 1224-1229	3.5	2
6	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
5	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
4	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses		2
3	Synchronous and Metachronous Breast and Ovarian Cancer: Experience From Two Large Cancer Center. *Frontiers in Oncology*, **2020**, 10, 608783	5.3	2
2	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. *Genetics in Medicine*, **2021**, 23, 1726-1737	8.1	2
1	Myasthenic syndrome and oligoclonal lymphocytosis: evolution into chronic lymphocytic leukemia. *Annals of Hematology*, **1998**, 76, 45-7	3	