

# Maria Adelaide Caligo

## List of Publications by Year in descending order

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158  
papers

8,496  
citations

53660

45  
h-index

54797

84  
g-index

166  
all docs

166  
docs citations

166  
times ranked

10882  
citing authors

#	ARTICLE	IF	CITATIONS
1	Pathology of Breast and Ovarian Cancers among <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers: Results from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 134-147.	1.1	513
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	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017, 49, 680-691.	9.4	356
4	Multicenter Comparative Multimodality Surveillance of Women at Genetic-Familial High Risk for Breast Cancer (HIBCRI Study): Interim Results. <i>Radiology</i> , 2007, 242, 698-715.	3.6	324
5	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
6	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	9.4	289
7	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. <i>Nature Genetics</i> , 2020, 52, 572-581.	9.4	265
8	Common Breast Cancer-Predisposition Alleles Are Associated with Breast Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>American Journal of Human Genetics</i> , 2008, 82, 937-948.	2.6	257
9	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
10	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2017, 109, .	3.0	242
11	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
12	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015, 47, 164-171.	9.4	221
13	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
14	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
15	Haplotype and Phenotype Analysis of Nine Recurrent <i>BRCA2</i> Mutations in 111 Families: Results of an International Study. <i>American Journal of Human Genetics</i> , 1998, 62, 1381-1388.	2.6	150
16	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
17	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
18	Germline mutations of the <i>BRCA1</i> -associated ring domain ( <i>BARD1</i> ) gene in breast and breast/ovarian families negative for <i>BRCA1</i> and <i>BRCA2</i> alterations. <i>Genes Chromosomes and Cancer</i> , 2002, 33, 235-242.	1.5	106

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19	Current policies for surveillance and management in women at risk of breast and ovarian cancer: a survey among 16 European family cancer clinics. <i>European Journal of Cancer</i> , 1998, 34, 1922-1926.	1.3	105
20	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
21	The Exon 13 Duplication in the <i>BRCA1</i> Gene Is a Founder Mutation Present in Geographically Diverse Populations. <i>American Journal of Human Genetics</i> , 2000, 67, 207-212.	2.6	100
22	Common variants in <i>LSP1</i> , 2q35 and 8q24 and breast cancer risk for <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>Human Molecular Genetics</i> , 2009, 18, 4442-4456.	1.4	99
23	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
24	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	5.8	93
25	Interplay between <i>BRCA1</i> and <i>RHAMM</i> Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. <i>PLoS Biology</i> , 2011, 9, e1001199.	2.6	91
26	<i>FANCM</i> c.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. <i>Human Molecular Genetics</i> , 2015, 24, 5345-5355.	1.4	91
27	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
28	Male breast cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: pathology data from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2016, 18, 15.	2.2	88
29	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019, 10, 431.	5.8	88
30	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
31	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in <i>ZNF365</i> are associated with breast cancer risk for <i>BRCA1</i> and/or <i>BRCA2</i> mutation carriers. <i>Breast Cancer Research</i> , 2012, 14, R33.	2.2	78
32	Common breast cancer susceptibility alleles are associated with tumour subtypes in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers: results from the Consortium of Investigators of Modifiers of <i>BRCA1/2</i> . <i>Breast Cancer Research</i> , 2011, 13, R110.	2.2	71
33	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>Human Molecular Genetics</i> , 2011, 20, 3304-3321.	1.4	68
34	<i>NM23</i> gene expression correlates with cell growth rate and S-phase. <i>International Journal of Cancer</i> , 1995, 60, 837-842.	2.3	66
35	Aberrant expression of <i>BARD1</i> in breast and ovarian cancers with poor prognosis. <i>International Journal of Cancer</i> , 2006, 118, 1215-1226.	2.3	63
36	Microsatellite instability and mismatch repair gene inactivation in sporadic pancreatic and colon tumours. <i>British Journal of Cancer</i> , 1999, 80, 11-16.	2.9	60

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37	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2014, 16, 3416.	2.2	57
38	A Novel Breast Cancer-Associated <i>BRIP1</i> ( <i>FANCD1/BACH1</i> ) Germ-line Mutation Impairs Protein Stability and Function. <i>Clinical Cancer Research</i> , 2008, 14, 4672-4680.	3.2	56
39	Evaluation of widely used models for predicting BRCA1 and BRCA2 mutations. <i>Journal of Medical Genetics</i> , 2004, 41, 278-285.	1.5	55
40	Penetrances of breast and ovarian cancer in a large series of families tested for BRCA1/2 mutations. <i>European Journal of Human Genetics</i> , 2004, 12, 899-906.	1.4	55
41	Stimulation of carrot somatic embryogenesis by proline and serine. <i>Plant Cell Reports</i> , 1984, 3, 210-214.	2.8	54
42	A Transcriptome-Wide Association Study Among 97,898 Women to Identify Candidate Susceptibility Genes for Epithelial Ovarian Cancer Risk. <i>Cancer Research</i> , 2018, 78, 5419-5430.	0.4	54
43	BRCA1 and BRCA2 germline mutations in Moroccan breast/ovarian cancer families: Novel mutations and unclassified variants. <i>Gynecologic Oncology</i> , 2012, 125, 687-692.	0.6	53
44	Thenm23 gene maps to human chromosome band 17q22 and shows a restriction fragment length polymorphism with <i>bg/III</i> . <i>Genes Chromosomes and Cancer</i> , 1992, 4, 84-88.	1.5	52
45	Cyclin D1 Overexpression in Thyroid Carcinomas: Relation with Clinico-Pathological Parameters, Retinoblastoma Gene Product, and Ki67 Labeling Index. <i>Thyroid</i> , 2000, 10, 741-746.	2.4	50
46	Down-regulation of thenm23.h1 gene inhibits cell proliferation. , 1997, 73, 297-302.		49
47	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
48	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. <i>Clinical Cancer Research</i> , 2011, 17, 3742-3750.	3.2	47
49	Common Variants at the 19p13.1 and <i>ZNF365</i> Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in <i>BRCA1</i> and <i>BRCA2</i> Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 645-657.	1.1	47
50	RNA-based analysis of BRCA1 and BRCA2 gene alterations. <i>Cancer Genetics and Cytogenetics</i> , 2006, 170, 93-101.	1.0	46
51	Usefulness of Technetium-99m Hexamethylpropylene Amine Oxime-Labeled Leukocyte Scintigraphy to Detect Pancreatic Necrosis in Patients with Acute Pancreatitis. <i>Pancreatology</i> , 2007, 7, 459-469.	0.5	46
52	Genetic alterations in hereditary breast cancer. <i>Annals of Oncology</i> , 2004, 15, i7-i13.	0.6	45
53	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2011, 103, 105-116.	3.0	40
54	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. <i>Human Mutation</i> , 2004, 24, 100-101.	1.1	39

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55	A yeast recombination assay to characterize human <i>BRCA1</i> missense variants of unknown pathological significance. <i>Human Mutation</i> , 2009, 30, 123-133.	1.1	39
56	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
57	<i>BRCA1</i> germline mutational spectrum in Italian families from Tuscany: a high frequency of novel mutations. <i>Oncogene</i> , 1996, 13, 1483-8.	2.6	37
58	Down regulation of NM23.H1, NM23.H2 and c-myc genes during differentiation induced by 1,25 dihydroxyvitamin D3. <i>Leukemia Research</i> , 1996, 20, 161-167.	0.4	36
59	High level of messenger RNA for <i>BRMS1</i> in primary breast carcinomas is associated with poor prognosis. <i>International Journal of Cancer</i> , 2007, 120, 1169-1178.	2.3	35
60	<i>PIK3CA</i> in Breast Carcinoma. <i>Diagnostic Molecular Pathology</i> , 2009, 18, 200-205.	2.1	34
61	Ovarian cancer susceptibility alleles and risk of ovarian cancer in <i>BRCA1</i> and <i>BRCA2</i> mutation carriers. <i>Human Mutation</i> , 2012, 33, 690-702.	1.1	34
62	Common variants of the <i>BRCA1</i> wild-type allele modify the risk of breast cancer in <i>BRCA1</i> mutation carriers. <i>Human Molecular Genetics</i> , 2011, 20, 4732-4747.	1.4	32
63	Transcriptome-wide association study of breast cancer risk by estrogen receptor status. <i>Genetic Epidemiology</i> , 2020, 44, 442-468.	0.6	32
64	<i>PALB2</i> : a novel inactivating mutation in a Italian breast cancer family. <i>Familial Cancer</i> , 2010, 9, 531-536.	0.9	30
65	Height and Body Mass Index as Modifiers of Breast Cancer Risk in <i>BRCA1/2</i> Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019, 111, 350-364.	3.0	30
66	Association between the <i>BRCA2</i> N372H variant and male breast cancer risk: a population-based case-control study in Tuscany, Central Italy. <i>BMC Cancer</i> , 2007, 7, 170.	1.1	28
67	The <i>FANCM</i> :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019, 5, 38.	2.3	28
68	Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian <i>BRCA1/2</i> carriers. <i>Breast Cancer Research and Treatment</i> , 2007, 103, 29-36.	1.1	27
69	<i>NM23</i> gene expression in human breast carcinomas: Loss of correlation with cell proliferation in the advanced phase of tumor progression. , 1997, 74, 102-111.		26
70	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in <i>BRCA2</i> mutation carriers. <i>Breast Cancer Research</i> , 2015, 17, 61.	2.2	26
71	Evidence for <i>SMAD3</i> as a modifier of breast cancer risk in <i>BRCA2</i> mutation carriers. <i>Breast Cancer Research</i> , 2010, 12, R102.	2.2	25
72	Germline investigation in male breast cancer of DNA repair genes by next-generation sequencing. <i>Breast Cancer Research and Treatment</i> , 2019, 178, 557-564.	1.1	24

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73	A low NM23.H1 gene expression identifying high malignancy human melanomas. <i>Melanoma Research</i> , 1994, 4, 179-184.	0.6	23
74	Ethical, Social and Economic Issues in Familial Breast Cancer: A Compilation of Views from the E.C. Biomed II Demonstration Project. <i>Disease Markers</i> , 1999, 15, 125-131.	0.6	23
75	Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , 2011, 13, R40.	2.2	23
76	A Nonsynonymous Polymorphism in <i>IRS1</i> Modifies Risk of Developing Breast and Ovarian Cancers in <i>BRCA1</i> and Ovarian Cancer in <i>BRCA2</i> Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1362-1370.	1.1	23
77	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. <i>European Journal of Human Genetics</i> , 2022, 30, 349-362.	1.4	23
78	Different Expressivity of BRCA1 and BRCA2: Analysis of 179 Italian Pedigrees with Identified Mutation. <i>Breast Cancer Research and Treatment</i> , 2003, 81, 71-79.	1.1	22
79	Reconstructing the Genealogy of a BRCA1 Founder Mutation by Phylogenetic Analysis. <i>Annals of Human Genetics</i> , 2008, 72, 310-318.	0.3	22
80	Effects of BRCA2 cis-regulation in normal breast and cancer risk amongst BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012, 14, R63.	2.2	22
81	Two mutations of BRCA2 gene at exon and splicing site in a woman who underwent oncogenetic counseling. <i>Annals of Oncology</i> , 2009, 20, 874-878.	0.6	21
82	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. <i>Breast</i> , 2013, 22, 1130-1135.	0.9	21
83	Lung Function Longitudinal Study by Phenotype and Genotype in Primary Ciliary Dyskinesia. <i>Chest</i> , 2020, 158, 117-120.	0.4	20
84	Identification of novel alternatively spliced BRCA1-associated RING domain (BARD1) messenger RNAs in human peripheral blood lymphocytes and in sporadic breast cancer tissues. <i>Genes Chromosomes and Cancer</i> , 2007, 46, 791-795.	1.5	19
85	Effect of the expression of BRCA2 on spontaneous homologous recombination and DNA damage-induced nuclear foci in <i>Saccharomyces cerevisiae</i> . <i>Mutagenesis</i> , 2013, 28, 187-195.	1.0	19
86	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and) Tumor Tissue for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. <i>JCO Precision Oncology</i> , 2018, 2, 1-42.	1.5	19
87	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. <i>British Journal of Cancer</i> , 2019, 121, 180-192.	2.9	19
88	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , 2021, 12, 1078.	5.8	19
89	Breast and Prostate Cancer Risks for Male <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variant Carriers Using Polygenic Risk Scores. <i>Journal of the National Cancer Institute</i> , 2022, 114, 109-122.	3.0	19
90	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. <i>Human Genetics</i> , 2011, 130, 685-699.	1.8	18

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91	MSH2 role in BRCA1-driven tumorigenesis: A preliminary study in yeast and in human tumors from BRCA1-VUS carriers. <i>European Journal of Medical Genetics</i> , 2015, 58, 531-539.	0.7	18
92	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016, 141, 386-401.	0.6	18
93	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
94	Functional Interaction Between BRCA1 and DNA Repair in Yeast May Uncover a Role of RAD50, RAD51, MRE11A, and MSH6 Somatic Variants in Cancer Development. <i>Frontiers in Genetics</i> , 2018, 9, 397.	1.1	18
95	Whole exome sequencing in familial isolated primary hyperparathyroidism. <i>Journal of Endocrinological Investigation</i> , 2020, 43, 231-245.	1.8	18
96	Haplotype analysis of BRCA1 gene reveals a new gene rearrangement: characterization of a 19.9 KBP deletion. <i>European Journal of Human Genetics</i> , 2004, 12, 775-777.	1.4	17
97	Mouse mammary tumor virus (MMTV) - like exogenous sequences are associated with sporadic but not hereditary human breast carcinoma. <i>Aging</i> , 2019, 11, 7236-7241.	1.4	17
98	Proline and serine affect polarity and development of carrot somatic embryos. <i>Cell Differentiation</i> , 1985, 17, 193-198.	1.3	16
99	p53 Inactivation is a Rare Event in Familial Breast Tumors Negative for BRCA1 and BRCA2 Mutations. <i>Breast Cancer Research and Treatment</i> , 2003, 82, 1-9.	1.1	16
100	Multimodal Assessment of Protein Functional Deficiency Supports Pathogenicity of BRCA1 p.V1688del. <i>Cancer Research</i> , 2009, 69, 7030-7037.	0.4	16
101	Common Genetic Variation at BARD1 Is Not Associated with Breast Cancer Risk in BRCA1 or BRCA2 Mutation Carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011, 20, 1032-1038.	1.1	16
102	Characterization of three alternative transcripts of the BRCA1 gene in patients with breast cancer and a family history of breast and/or ovarian cancer who tested negative for pathogenic mutations. <i>International Journal of Molecular Medicine</i> , 2015, 35, 950-956.	1.8	16
103	Germline mutations in DNA repair genes may predict neoadjuvant therapy response in triple negative breast patients. <i>Genes Chromosomes and Cancer</i> , 2016, 55, 915-924.	1.5	16
104	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. <i>Genetics in Medicine</i> , 2021, 23, 1726-1737.	1.1	16
105	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). <i>British Journal of Cancer</i> , 2009, 101, 2048-2054.	2.9	15
106	BRCA1 and BRCA2 5' noncoding region variants identified in breast cancer patients alter promoter activity and protein binding. <i>Human Mutation</i> , 2018, 39, 2025-2039.	1.1	15
107	PROMs in post-mastectomy care: Patient self-reports (BREAST-Q) as a powerful instrument to personalize medical services. <i>European Journal of Surgical Oncology</i> , 2020, 46, 1034-1040.	0.5	15
108	Decreasing expression of NM23 gene in metastatic murine mammary tumors of viral etiology (MMTV). <i>Anticancer Research</i> , 1992, 12, 969-73.	0.5	15

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109	Defective interleukin six expression and responsiveness in human mammary cells transformed by an adeno 5/SV40 hybrid virus. <i>British Journal of Cancer</i> , 1996, 73, 1356-1361.	2.9	14
110	Microsatellite alterations and p53, TGF $\beta$ RII, IGF1R and BAX mutations in sporadic non-small-cell lung cancer. , 1998, 78, 606-609.		13
111	Insurance Implications for Individuals with a High Risk of Breast and Ovarian Cancer in Europe. <i>Disease Markers</i> , 1999, 15, 159-165.	0.6	13
112	Effect of the overexpression of BRCA2 unclassified missense variants on spontaneous homologous recombination in human cells. <i>Breast Cancer Research and Treatment</i> , 2011, 129, 1001-1009.	1.1	13
113	Microsatellite alterations and K-ras, TGF $\beta$ RII, IGF1R and bax mutations in sporadic cancers of the gastrointestinal tract.. <i>Oncology Reports</i> , 2000, 7, 1371-5.	1.2	13
114	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. <i>Breast Cancer Research and Treatment</i> , 2009, 117, 371-379.	1.1	12
115	Identification of BRAF 3' UTR Isoforms in Melanoma. <i>Journal of Investigative Dermatology</i> , 2015, 135, 1694-1697.	0.3	12
116	Carcinosarcoma of the Breast: An Aggressive Subtype of Metaplastic Cancer. Report of a Rare Case in a Young BRCA-1 Mutated Woman. <i>Clinical Breast Cancer</i> , 2017, 17, e31-e35.	1.1	12
117	Next generation sequencing technologies for a successful diagnosis in a cold case of Leigh syndrome. <i>BMC Neurology</i> , 2018, 18, 99.	0.8	12
118	Effect of BRCA1 missense variants on gene reversion in DNA double-strand break repair mutants and cell cycle-arrested cells of <i>Saccharomyces cerevisiae</i> . <i>Mutagenesis</i> , 2020, 35, 189-195.	1.0	12
119	Longitudinal Lung Volume Changes by Ultrastructure and Genotype in Primary Ciliary Dyskinesia. <i>Annals of the American Thoracic Society</i> , 2021, 18, 963-970.	1.5	12
120	Origin of araC-induced endoreduplicated cells. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 1987, 177, 261-265.	0.4	11
121	Breast Cancer Risk and 6q22.33: Combined Results from Breast Cancer Association Consortium and Consortium of Investigators on Modifiers of BRCA1/2. <i>PLoS ONE</i> , 2012, 7, e35706.	1.1	11
122	Disorders of sexual development with XY karyotype and female phenotype: clinical findings and genetic background in a cohort from a single centre. <i>Journal of Endocrinological Investigation</i> , 2021, 44, 145-151.	1.8	11
123	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , 2016, 11, e0158801.	1.1	10
124	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
125	Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome: New Report with a 197-kb Deletion Upstream of FOXL2 and Review of the Literature. <i>Molecular Syndromology</i> , 2019, 10, 147-153.	0.3	9
126	Genomic PCR-SSCP analysis of the metastasis associated NM23-H1 (NME1) gene: a study on colorectal cancer. <i>Anticancer Research</i> , 1993, 13, 2149-54.	0.5	9



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127	Increased rate of base substitution in a hamster mutator strain obtained during serial selection for gene amplification.. <i>Molecular and Cellular Biology</i> , 1990, 10, 6805-6808.	1.1	8
128	Risk Estimation as a Decision-Making Tool for Genetic Analysis of the Breast Cancer Susceptibility Genes. <i>Disease Markers</i> , 1999, 15, 53-65.	0.6	8
129	The KL-VS sequence variant of Klotho and cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2012, 132, 1119-1126.	1.1	8
130	Expression of human poly (ADP-ribose) polymerase 1 in <i>Saccharomyces cerevisiae</i> : Effect on survival, homologous recombination and identification of genes involved in intracellular localization. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2015, 774, 14-24.	0.4	8
131	Whole-exome analysis of a Liê“Fraumeni family trio with a novel TP53 PRD mutation and anticipation profile. <i>Carcinogenesis</i> , 2017, 38, 938-943.	1.3	8
132	Time course of sister chromatid exchanges and gene amplification induced by 1-?-d-arabinofuranosylcytosine in V79-AP4 Chinese hamster cells. <i>Chromosoma</i> , 1988, 96, 306-310.	1.0	7
133	Clinicopathological Significance of GADD45 Gene Alterations in Human Familial Breast Carcinoma. <i>Breast Cancer Research and Treatment</i> , 2004, 87, 197-201.	1.1	7
134	Evaluation of the XRCC1 gene as a phenotypic modifier in BRCA1/2 mutation carriers. Results from the consortium of investigators of modifiers of BRCA1/BRCA2. <i>British Journal of Cancer</i> , 2011, 104, 1356-1361.	2.9	7
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