

Maria Adelaide Caligo

List of Publications by Citations

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158
papers

6,303
citations

44
h-index

75
g-index

166
ext. papers

7,588
ext. citations

7.1
avg, IF

4.17
L-index

#	Paper	IF	Citations
158	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 134-47	4	411
157	Multicenter comparative multimodality surveillance of women at genetic-familial high risk for breast cancer (HIBCRIIT study): interim results. <i>Radiology</i> , 2007 , 242, 698-715	20.5	287
156	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. <i>JAMA - Journal of the American Medical Association</i> , 2015 , 313, 1347-61	27.4	286
155	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. <i>Nature Genetics</i> , 2010 , 42, 885-92	36.3	276
154	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>American Journal of Human Genetics</i> , 2008 , 82, 937-48	11	218
153	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. <i>PLoS Genetics</i> , 2013 , 9, e1003212	6	209
152	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , 2017 , 49, 680-691	36.3	190
151	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017 , 49, 1767-1778	36.3	186
150	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , 2015 , 47, 164-71	36.3	177
149	Evaluation of Polygenic Risk Scores for Breast and Ovarian Cancer Risk Prediction in BRCA1 and BRCA2 Mutation Carriers. <i>Journal of the National Cancer Institute</i> , 2017 , 109,	9.7	153
148	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. <i>Cancer Research</i> , 2010 , 70, 9742-54	10.1	147
147	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , 2018 , 39, 593-620	4.7	138
146	Haplotype and phenotype analysis of nine recurrent BRCA2 mutations in 111 families: results of an international study. <i>American Journal of Human Genetics</i> , 1998 , 62, 1381-8	11	138
145	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. <i>Journal of Clinical Oncology</i> , 2017 , 35, 2240-2250	2.2	101
144	Current policies for surveillance and management in women at risk of breast and ovarian cancer: a survey among 16 European family cancer clinics. European Familial Breast Cancer Collaborative Group. <i>European Journal of Cancer</i> , 1998 , 34, 1922-6	7.5	98
143	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016 , 48, 374-86	36.3	93
142	Germline mutations of the BRCA1-associated ring domain (BARD1) gene in breast and breast/ovarian families negative for BRCA1 and BRCA2 alterations. <i>Genes Chromosomes and Cancer</i> , 2002 , 33, 235-42	5	93

141	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2009 , 18, 4442-56	5.6	91
140	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. <i>American Journal of Human Genetics</i> , 2000 , 67, 207-212	11	89
139	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	36.3	76
138	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. <i>PLoS Biology</i> , 2011 , 9, e1001199	9.7	73
137	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012 , 14, R33	8.3	70
136	FANCM 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, 5345-55	5.6	68
135	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016 , 7, 11375	17.4	64
134	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. <i>Breast Cancer Research</i> , 2011 , 13, R110	8.3	62
133	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2011 , 20, 3304-21	5.6	62
132	NM23 gene expression correlates with cell growth rate and S-phase. <i>International Journal of Cancer</i> , 1995 , 60, 837-42	7.5	60
131	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , 2016 , 18, 15	8.3	58
130	Microsatellite instability and mismatch repair gene inactivation in sporadic pancreatic and colon tumours. <i>British Journal of Cancer</i> , 1999 , 80, 11-6	8.7	56
129	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020 , 52, 56-73	36.3	56
128	Aberrant expression of BARD1 in breast and ovarian cancers with poor prognosis. <i>International Journal of Cancer</i> , 2006 , 118, 1215-26	7.5	55
127	Comparison of mRNA splicing assay protocols across multiple laboratories: recommendations for best practice in standardized clinical testing. <i>Clinical Chemistry</i> , 2014 , 60, 341-52	5.5	53
126	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , 2019 , 40, 1557-1578	4.7	52
125	Evaluation of widely used models for predicting BRCA1 and BRCA2 mutations. <i>Journal of Medical Genetics</i> , 2004 , 41, 278-85	5.8	50
124	Stimulation of carrot somatic embryogenesis by proline and serine. <i>Plant Cell Reports</i> , 1984 , 3, 210-4	5.1	50

123	The NM23 gene maps to human chromosome band 17q22 and shows a restriction fragment length polymorphism with BglII. <i>Genes Chromosomes and Cancer</i> , 1992 , 4, 84-8	5	49
122	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. <i>Nature Communications</i> , 2019 , 10, 1741	17.4	47
121	BRCA1 and BRCA2 germline mutations in Moroccan breast/ovarian cancer families: novel mutations and unclassified variants. <i>Gynecologic Oncology</i> , 2012 , 125, 687-92	4.9	46
120	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	8.3	46
119	A novel breast cancer-associated BRIP1 (FANCI/BACH1) germ-line mutation impairs protein stability and function. <i>Clinical Cancer Research</i> , 2008 , 14, 4672-80	12.9	46
118	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	5.3	46
117	Shared heritability and functional enrichment across six solid cancers. <i>Nature Communications</i> , 2019 , 10, 431	17.4	45
116	The role of KRAS rs61764370 in invasive epithelial ovarian cancer: implications for clinical testing. <i>Clinical Cancer Research</i> , 2011 , 17, 3742-50	12.9	45
115	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. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 645-57	4	44
114	Cyclin D1 overexpression in thyroid carcinomas: relation with clinico-pathological parameters, retinoblastoma gene product, and Ki67 labeling index. <i>Thyroid</i> , 2000 , 10, 741-6	6.2	44
113	Down-regulation of the nm23.h1 gene inhibits cell proliferation. <i>International Journal of Cancer</i> , 1997 , 73, 297-302	7.5	40
112	RNA-based analysis of BRCA1 and BRCA2 gene alterations. <i>Cancer Genetics and Cytogenetics</i> , 2006 , 170, 93-101		40
111	Cancer risk among the relatives of patients with pancreatic ductal adenocarcinoma. <i>Pancreatology</i> , 2007 , 7, 459-69	3.8	39
110	Genetic alterations in hereditary breast cancer. <i>Annals of Oncology</i> , 2004 , 15 Suppl 1, I7-I13	10.3	39
109	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, 105-16	9.7	37
108	The CHEK2 c.1100delC mutation plays an irrelevant role in breast cancer predisposition in Italy. <i>Human Mutation</i> , 2004 , 24, 100-1	4.7	36
107	BRCA1 germline mutational spectrum in Italian families from Tuscany: a high frequency of novel mutations. <i>Oncogene</i> , 1996 , 13, 1483-8	9.2	36
106	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , 2020 , 22, 1653-1666	8.1	34

105	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	10.1	32
104	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, 161-7	2.7	32
103	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Human Mutation</i> , 2012 , 33, 690-702	4.7	31
102	PIK3CA in breast carcinoma: a mutational analysis of sporadic and hereditary cases. <i>Diagnostic Molecular Pathology</i> , 2009 , 18, 200-5		31
101	A yeast recombination assay to characterize human BRCA1 missense variants of unknown pathological significance. <i>Human Mutation</i> , 2009 , 30, 123-33	4.7	30
100	PALB2: a novel inactivating mutation in a Italian breast cancer family. <i>Familial Cancer</i> , 2010 , 9, 531-6	3	30
99	High level of messenger RNA for BRMS1 in primary breast carcinomas is associated with poor prognosis. <i>International Journal of Cancer</i> , 2007 , 120, 1169-78	7.5	28
98	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). <i>JAMA Oncology</i> , 2020 , 6, 1218-1230	13.4	25
97	NM23 gene expression in human breast carcinomas: loss of correlation with cell proliferation in the advanced phase of tumor progression. <i>International Journal of Cancer</i> , 1997 , 74, 102-11	7.5	23
96	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, 125-31	3.2	23
95	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , 2020 , 80, 624-638	10.1	22
94	Methyl group metabolism gene polymorphisms as modifier of breast cancer risk in Italian BRCA1/2 carriers. <i>Breast Cancer Research and Treatment</i> , 2007 , 103, 29-36	4.4	22
93	Different expressivity of BRCA1 and BRCA2: analysis of 179 Italian pedigrees with identified mutation. <i>Breast Cancer Research and Treatment</i> , 2003 , 81, 71-9	4.4	22
92	Height and Body Mass Index as Modifiers of Breast Cancer Risk in BRCA1/2 Mutation Carriers: A Mendelian Randomization Study. <i>Journal of the National Cancer Institute</i> , 2019 , 111, 350-364	9.7	22
91	Evidence for SMAD3 as a modifier of breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2010 , 12, R102	8.3	21
90	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. <i>Human Molecular Genetics</i> , 2011 , 20, 4732-47	5.6	21
89	Association between the BRCA2 N372H variant and male breast cancer risk: a population-based case-control study in Tuscany, Central Italy. <i>BMC Cancer</i> , 2007 , 7, 170	4.8	21
88	A low NM23.H1 gene expression identifying high malignancy human melanomas. <i>Melanoma Research</i> , 1994 , 4, 179-84	3.3	21

87	Two mutations of BRCA2 gene at exon and splicing site in a woman who underwent oncogenetic counseling. <i>Annals of Oncology</i> , 2009 , 20, 874-8	10.3	20
86	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012 , 21, 1362-70	4	20
85	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	4.4	18
84	Reconstructing the genealogy of a BRCA1 founder mutation by phylogenetic analysis. <i>Annals of Human Genetics</i> , 2008 , 72, 310-8	2.2	17
83	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, 1130-5	3.6	16
82	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2015 , 17, 61	8.3	16
81	Exploring the link between MORF4L1 and risk of breast cancer. <i>Breast Cancer Research</i> , 2011 , 13, R40	8.3	16
80	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , 2016 , 141, 386-401	4.9	15
79	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	4.4	15
78	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, 950-6	4.4	15
77	Effects of BRCA2 cis-regulation in normal breast and cancer risk amongst BRCA2 mutation carriers. <i>Breast Cancer Research</i> , 2012 , 14, R63	8.3	15
76	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, 187-95	2.8	15
75	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. <i>Human Genetics</i> , 2011 , 130, 685-99	6.3	15
74	Multimodal assessment of protein functional deficiency supports pathogenicity of BRCA1 p.V1688del. <i>Cancer Research</i> , 2009 , 69, 7030-7	10.1	15
73	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, 775-7	5.3	15
72	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	4.4	14
71	Proline and serine affect polarity and development of carrot somatic embryos. <i>Cell Differentiation</i> , 1985 , 17, 193-198		14
70	Decreasing expression of NM23 gene in metastatic murine mammary tumors of viral etiology (MMTV). <i>Anticancer Research</i> , 1992 , 12, 969-73	2.3	14

69	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	8.7	13
68	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, 531-9	2.6	13
67	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, 2048-54	8.7	13
66	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, 1032-8	4	13
65	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	5	13
64	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. <i>Npj Breast Cancer</i> , 2019 , 5, 38	7.8	12
63	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, 1001-9	4.4	12
62	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, 1356-61	8.7	12
61	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and Breast/Ovarian) Cancer Susceptibility Genes: An International Survey by the Evidence-Based Network for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. <i>JCO Precision Oncology</i> , 2018 , 2,	3.6	12
60	Functional Interaction Between and DNA Repair in Yeast May Uncover a Role of , and Somatic Variants in Cancer Development. <i>Frontiers in Genetics</i> , 2018 , 9, 397	4.5	12
59	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	4.7	12
58	Identification of BRAF 3' UTR Isoforms in Melanoma. <i>Journal of Investigative Dermatology</i> , 2015 , 135, 1694-1697	4.3	11
57	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	5.6	11
56	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	3.7	10
55	No evidence that GATA3 rs570613 SNP modifies breast cancer risk. <i>Breast Cancer Research and Treatment</i> , 2009 , 117, 371-9	4.4	10
54	Microsatellite alterations and p53, TGFbetaRII, IGFIR and BAX mutations in sporadic non-small-cell lung cancer. <i>International Journal of Cancer</i> , 1998 , 78, 606-9	7.5	10
53	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, 791-5	5	10
52	Insurance implications for individuals with a high risk of breast and ovarian cancer in Europe. <i>Disease Markers</i> , 1999 , 15, 159-65	3.2	10

51	Origin of araC-induced endoreduplicated cells. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 1987 , 177, 261-5	3.3	10
50	Microsatellite alterations and K-ras, TGFbetaRII, IGFRII and bax mutations in sporadic cancers of the gastrointestinal tract. <i>Oncology Reports</i> , 2000 , 7, 1371-5	3.5	10
49	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. <i>Genetic Epidemiology</i> , 2020 , 44, 442-468	2.6	9
48	Next generation sequencing technologies for a successful diagnosis in a cold case of Leigh syndrome. <i>BMC Neurology</i> , 2018 , 18, 99	3.1	9
47	Whole exome sequencing in familial isolated primary hyperparathyroidism. <i>Journal of Endocrinological Investigation</i> , 2020 , 43, 231-245	5.2	9
46	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, 6805-8	4.8	8
45	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	2.3	8
44	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	3.6	8
43	Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome: New Report with a 197-kb Deletion Upstream of and Review of the Literature. <i>Molecular Syndromology</i> , 2019 , 10, 147-153	1.5	7
42	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	3.3	7
41	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, 1119-26	4.4	7
40	Risk estimation as a decision-making tool for genetic analysis of the breast cancer susceptibility genes. EC Demonstration Project on Familial Breast Cancer. <i>Disease Markers</i> , 1999 , 15, 53-65	3.2	7
39	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	3.7	7
38	Whole-exome analysis of a Li-Fraumeni family trio with a novel TP53 PRD mutation and anticipation profile. <i>Carcinogenesis</i> , 2017 , 38, 938-943	4.6	6
37	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, 1356-61	8.7	6
36	A recombination-based method to characterize human BRCA1 missense variants. <i>Breast Cancer Research and Treatment</i> , 2011 , 125, 265-72	4.4	6
35	Time course of sister chromatid exchanges and gene amplification induced by 1-beta-D-arabinofuranosylcytosine in V79-AP4 Chinese hamster cells. <i>Chromosoma</i> , 1988 , 96, 306-10	2.8	6
34	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	5.2	6

33	Lung Function Longitudinal Study by Phenotype and Genotype in Primary Ciliary Dyskinesia. <i>Chest</i> , 2020 , 158, 117-120	5.3	5
32	Choroid plexus carcinoma: a new case associated with a novel TP53 germ line mutation. <i>Neuropathology and Applied Neurobiology</i> , 2008 , 34, 564-8	5.2	5
31	Clinicopathological significance of GADD45 gene alterations in human familial breast carcinoma. <i>Breast Cancer Research and Treatment</i> , 2004 , 87, 197-201	4.4	5
30	Mutational analysis of the NM23.H1 gene in human breast cancer. <i>Cancer Genetics and Cytogenetics</i> , 2000 , 121, 181-5		5
29	Myelodysplastic syndromes: advantages of a combined cytogenetic and molecular diagnostic workup. <i>Oncotarget</i> , 2017 , 8, 79188-79200	3.3	5
28	Paternity in 5 α -Reductase-2 Deficiency: Report of Two Brothers with Spontaneous or Assisted Fertility and Literature Review. <i>Sexual Development</i> , 2019 , 13, 55-59	1.6	4
27	Identification of two novel BRCA1-partner genes in the DNA double-strand break repair pathway. <i>Breast Cancer Research and Treatment</i> , 2013 , 141, 515-22	4.4	4
26	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	3	4
25	Association of SULT1A1 Arg \rightarrow His polymorphism with male breast cancer risk: results from a multicenter study in Italy. <i>Breast Cancer Research and Treatment</i> , 2014 , 148, 623-8	4.4	4
24	Feline immunodeficiency virus vector as a tool for preventative strategies against human breast cancer. <i>Veterinary Immunology and Immunopathology</i> , 2010 , 134, 132-7	2	4
23	Characterisation of gene expression profiles of yeast cells expressing BRCA1 missense variants. <i>European Journal of Cancer</i> , 2009 , 45, 2187-96	7.5	4
22	A region on the long arm of chromosome 16 is frequently deleted in metastatic node-negative breast cancer. <i>International Journal of Oncology</i> , 1998 , 13, 177-82	1	4
21	Genetic testing for breast cancer predisposition in 1999: which molecular strategy and which family criteria?. <i>Disease Markers</i> , 1999 , 15, 67-8	3.2	4
20	NM23.H1 loss of heterozygosity in human mammary carcinomas. Definition of a deletion map on chromosome 17q. <i>Annals of the New York Academy of Sciences</i> , 1993 , 698, 136-42	6.5	4
19	Accumulation of anchorage independent cells showing amplified genes (CAD) during the in vitro propagation of CHEF18 Chinese hamster cells. <i>Cell Proliferation</i> , 1993 , 26, 161-70	7.9	4
18	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	2.8	4
17	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. <i>Nature Communications</i> , 2021 , 12, 1078	17.4	4
16	Evaluation of chromosome 6p22 as a breast cancer risk modifier locus in a follow-up study of BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , 2012 , 136, 295-302	4.4	3

15	Effects on human transcriptome of mutated BRCA1 BRCT domain: a microarray study. <i>BMC Cancer</i> , 2012 , 12, 207	4.8	3
14	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	4.8	3
13	Longitudinal Lung Volume Changes by Ultrastructure and Genotype in Primary Ciliary Dyskinesia. <i>Annals of the American Thoracic Society</i> , 2021 , 18, 963-970	4.7	3
12	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> , 2021 ,	9.7	3
11	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> , 2021 ,	8.1	2
10	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
9	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses		2
8	Disorders/Differences of Sex Development Presenting in the Newborn With 46,XY Karyotype. <i>Frontiers in Pediatrics</i> , 2021 , 9, 627281	3.4	2
7	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	8.1	2
6	Mutation Analysis of BRCA1 and BRCA2 in Italian Hereditary and Sporadic Forms of Breast and Ovarian Cancers: Tumor Genotype-Phenotype Correlation in Breast Cancer BRCA-Mutation Carriers. <i>Disease Markers</i> , 1999 , 15, 101-102	3.2	1
5	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1
4	Detection of Germline Variants in 450 Breast/Ovarian Cancer Families with a Multi-Gene Panel Including Coding and Regulatory Regions. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	1
3	Nutrition, epigenetic markers and growth in preterm infants. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2021 , 34, 3963-3968	2	0
2	BRCA1 gene variant p.P142H associated with male breast cancer: a two-generation genealogic study and literature review. <i>Familial Cancer</i> , 2015 , 14, 515-9	3	
1	Reply to BRCA2 splice site mutations in an Italian breast/ovarian cancer family. <i>Annals of Oncology</i> , 2009 , 20, 1285-1286	10.3	