Irene Konstantopoulou

List of Publications by Year
in descending order

91	5,545	117625	85541
papers	citations	34 h-index	71 g-index

95	95	95	8527
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Inherited Mutations in 17 Breast Cancer Susceptibility Genes Among a Large Triple-Negative Breast Cancer Cohort Unselected for Family History of Breast Cancer. Journal of Clinical Oncology, 2015, 33, 304-311.	1.6	521
2	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	21.4	374
3	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
4	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptorâ€“negative breast cancer. Nature Genetics, 2011, 43, 1210-1214.	21.4	279
5	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
6	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.	2.5	269
7	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
8	Triple-Negative Breast Cancer Risk Genes Identified by Multigene Hereditary Cancer Panel Testing. Journal of the National Cancer Institute, 2018, 110, 855-862.	6.3	225
9	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
10	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
11	Insecticidal effects of essential oils. A study of the effects of essential oils extracted from eleven Greek aromatic plants onDrosophila auraria. Experientia, 1992, 48, 616-619.	1.2	152
12	Genome-wide association study identifies 25 known breast cancer susceptibility loci as risk factors for triple-negative breast cancer. Carcinogenesis, 2014, 35, 1012-1019.	2.8	145
13	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
14	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
15	Common Breast Cancer Susceptibility Loci Are Associated with Triple-Negative Breast Cancer. Cancer Research, 2011, 71, 6240-6249.	0.9	109
16	On the origin and diffusion of BRCA1 c.5266dupC (5382insC) in European populations. European Journal of Human Genetics, 2011, 19, 300-306.	2.8	107
17	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
18	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90

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19	Cancer Risks Associated With BRCA1 and BRCA2 Pathogenic Variants. Journal of Clinical Oncology, 2022, 40, 1529-1541.	1.6	90
20	Prevalence of BRCA1 mutations among 403 women with triple-negative breast cancer: implications for genetic screening selection criteria: a Hellenic Cooperative Oncology Group Study. Breast Cancer Research and Treatment, 2012, 134, 353-362.	2.5	82
21	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
22	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
23	Prevalence of GJB2 mutations in prelingual deafness in the Greek population. International Journal of Pediatric Otorhinolaryngology, 2002, 65, 101-108.	1.0	63
24	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.	5.0	57
25	Characterization of the Cancer Spectrum in Men With GermlineBRCA1andBRCA2Pathogenic Variants. JAMA Oncology, 2020, 6, 1218.	7.1	48
26	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
27	Novel genomic rearrangements in the BRCA1 gene detected in greek breast/ovarian cancer patients. European Journal of Cancer, 2007, 43, 443-453.	2.8	45
28	Germ line BRCA1 & BRCA2 mutations in Greek breast/ovarian cancer families: 5382insC is the most frequent mutation observed. Cancer Letters, 2002, 185, 61-70.	7.2	42
29	A rareRETgene exonÂ8 mutation is found in two Greek kindreds with familial medullary thyroid carcinoma: implications for screening. Clinical Endocrinology, 2006, 64, 561-566.	2.4	41
30	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
31	Greek BRCA1 and BRCA2 mutation spectrum: two BRCA1 mutations account for half the carriers found among high-risk breast/ovarian cancer patients. Breast Cancer Research and Treatment, 2008, 107, 431-441.	2.5	38
32	G1738R is a BRCA1 founder mutation in Greek breast/ovarian cancer patients: evaluation of its pathogenicity and inferences on its genealogical history. Breast Cancer Research and Treatment, 2008, 110, 377-385.	2.5	37
33	Association of the VariantsCASP8D302H andCASP10V410I with Breast and Ovarian Cancer Risk inBRCA1andBRCA2Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 2859-2868.	2.5	37
34	Germline deleterious mutations in genes other than BRCA2 are infrequent in male breast cancer. Breast Cancer Research and Treatment, 2018, 169, 105-113.	2.5	37
35	BRCA2 gene mutations in Greek patients with familial breast cancer. Human Mutation, 2002, 19, 81-82.	2.5	35
36	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

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37	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. Human Mutation, 2019, 40, e1-e23.	2.5	34
38	Functional characterization of CHEK2 variants in a Saccharomyces cerevisiae system. Human Mutation, 2019, 40, 631-648.	2.5	34
39	High prevalence of <scp>BRCA1</scp> founder mutations in Greek breast/ovarian families. Clinical Genetics, 2014, 85, 36-42.	2.0	33
40	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
41	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
42	A Drosophila hsp70 gene contains long, antiparallel, coupled open reading frames (LAC ORFs) conserved in homologous loci. Journal of Molecular Evolution, 1995, 41, 414-420.	1.8	28
43	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
44	BRCA1 mutation analysis in breast/ovarian cancer families from Greece. Human Mutation, 2000, 16, 272-273.	2.5	27
45	Prevalence of BRCA1 Mutations in Familial and Sporadic Greek Ovarian Cancer Cases. PLoS ONE, 2013, 8, e58182.	2.5	27
46	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.	5.0	26
47	Contribution of BRCA1 germ-line mutations to breast cancer in Greece: a hospital-based study of 987 unselected breast cancer cases. British Journal of Cancer, 2009, 101, 32-37.	6.4	24
48	The hsp70 locus of Drosophila auraria (montium subgroup) is single and contains copies in a conserved arrangement. Chromosoma, 1998, 107, 577-586.	2.2	23
49	Extending the clinical phenotype associated with biallelic NTHL1 germline mutations. Clinical Genetics, 2018, 94, 588-589.	2.0	23
50	Growth index is independent of microvessel density in nonâ€“small-cell lung carcinomas. Human Pathology, 2002, 33, 812-818.	2.0	21
51	The heat shock genes in theDrosophila montium subgroup: Chromosomal localization and evolutionary implications. Chromosoma, 1996, 105, 104-110.	2.2	20
52	A change in the last base of BRCA1 exon 23, 5586Gâ†'A, results in abnormal RNA splicing. Cancer Genetics and Cytogenetics, 2002, 134, 175-177.	1.0	19
53	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2009, 101, 1456-1460.	6.4	19
54	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18

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55	Haplotype analysis reveals that the recurrent BRCA1 deletion of exons 23 and 24 is a Greek founder mutation. Clinical Genetics, 2017, 91, 482-487.	2.0	18
56	The Heat-Shock Gene hsp83 of Drosophila auraria: Genomic Organization, Nucleotide Sequence, and Long Antiparallel Coupled ORFs (LAC ORFs). Journal of Molecular Evolution, 1998, 46, 334-343.	1.8	16
57	The predictive ability of the 313 variantâ€“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.	2.4	16
58	The Afrotropical Drosophila montium subgroup: Balbiani ring 1, polytene chromosomes, and heat shock response of Drosophila vulcana. Genome, 1996, 39, 588-597.	2.0	13
59	Prelingual Nonsyndromic Hearing Loss in Greece. Orl, 2002, 64, 321-323.	1.1	13
60	alphaB-crystallin is a marker of aggressive breast cancer behavior but does not independently predict for patient outcome: a combined analysis of two randomized studies. BMC Clinical Pathology, 2014, 14, 28.	1.8	13
61	Ichthyosis Follicularis with Alopecia and Photophobia in a Girl with Cataract: Histological and Electron Microscopy Findings. Acta Dermato-Venereologica, 2005, 85, 51-55.	1.3	12
62	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. Human Molecular Genetics, 2014, 23, 6034-6046.	2.9	12
63	Genetic analysis and clinical description of Greek patients with Peutz-Jeghers syndrome: Creation of a National Registry. Cancer Genetics, 2018, 220, 19-23.	0.4	12
64	Cross-Cancer Genome-Wide Association Study of Endometrial Cancer and Epithelial Ovarian Cancer Identifies Genetic Risk Regions Associated with Risk of Both Cancers. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 217-228.	2.5	12
65	Haplotype analysis of two recurrent genomic rearrangements in the BRCA1 gene suggests they are founder mutations for the Greek population. Clinical Genetics, 2011, 80, 375-382.	2.0	11
66	CHEK2 c.1100delC allele is rarely identified in Greek breast cancer cases. Cancer Genetics, 2015, 208, 129-134.	0.4	11
67	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	3.7	11
68	Pathology of BRCA1- and BRCA2-associated Breast Cancers: Known and Less Known Connections. Clinical Breast Cancer, 2020, 20, 152-159.	2.4	9
69	PALB2 c.2257C>T truncating variant is a Greek founder and is associated with high breast cancer risk. Journal of Human Genetics, 2019, 64, 767-773.	2.3	8
70	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. British Journal of Cancer, 2011, 104, 1356-1361.	6.4	7
71	Characterization and prevalence of two novel CHEK2 large deletions in Greek breast cancer patients. Journal of Human Genetics, 2018, 63, 877-886.	2.3	7
72	Contribution of RAD51D germline mutations in breast and ovarian cancer in Greece. Journal of Human Genetics, 2018, 63, 1149-1158.	2.3	7

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73	Prevalence and founder effect of the BRCA1 p.(Val1833Met) variant in the Greek population, with further evidence for pathogenicity and risk modification. Cancer Genetics, 2019, 237, 90-96.	0.4	7
74	BRCA1 and BRCA2 germline testing in Cretan isolates reveals novel and strong founder effects. International Journal of Cancer, 2020, 147, 1334-1342.	5.1	7
75	Lack of association between RNASEL Arg462Gln variant and the risk of breast cancer. Anticancer Research, 2004, 24, 2547-9.	1.1	7
76	Atypical Medullary Breast Carcinoma in a Family Carrying the 5382insC BRCA-1 Mutation. Breast Journal, 2003, 9, 260-262.	1.0	6
77	CHEK2 Pathogenic Variants in Greek Breast Cancer Patients: Evidence for Strong Associations with Estrogen Receptor Positivity, Overuse of Risk-Reducing Procedures and Population Founder Effects. Cancers, 2021, 13, 2106.	3.7	5
78	Variations in the heat-induced protein pattern of severalDrosophila montiumsubgroup species (Diptera: Drosophilidae). Genome, 1997, 40, 132-137.	2.0	4
79	Genetic evaluation based on family history and Her2 status correctly identifies <scp>TP53</scp> mutations in very early onset breast cancer cases. Clinical Genetics, 2015, 87, 383-387.	2.0	4
80	The fate of BRCA1-related germline mutations in triple-negative breast tumors. American Journal of Cancer Research, 2017, 7, 98-114.	1.4	4
81	A Paternally InheritedBRCA1Mutation Associated with an Unusual Aggressive Clinical Phenotype. Case Reports in Genetics, 2014, 2014, 1-3.	0.2	3
82	BRCA1 and BRCA2 genes mutation analysis in patients with a family history of breast and ovarian cancer. Journal of Medical Biochemistry, 2004, 23, 271-277.	0.1	3
83	The heat shock genes in the Drosophila montium subgroup: chromosomal localization and evolutionary implications. Chromosoma, 1996, 105, 104-110.	2.2	2
84	Lung cancer as a predominant feature in a patient with <scp>Peutzâ€“Jeghers</scp> syndrome: Case report. Thoracic Cancer, 2022, , .	1.9	2
85	A Patient Affected with Serous Ovarian/Peritoneal Carcinoma Carrying the FANCM Mutation. Case Reports in Oncological Medicine, 2019, 2019, 1-2.	0.3	1
86	CanVaS: Documenting the genetic variation spectrum of Greek cancer patients. Human Mutation, 2021, 42, 1081-1093.	2.5	1
87	When cascade testing for familial variant seems inadequate to provide clinically actionable information for blood relatives. Cancer Genetics, 2021, 258-259, 49-50.	0.4	1
88	Pathogenic Variants in Breast Cancer Susceptibility Genes in Older Women. JAMA - Journal of the American Medical Association, 2020, 324, 397.	7.4	0
89	Variant Interpretation in Patients With Metastatic Breast Cancer. JAMA Oncology, 2020, 6, 581.	7.1	0
90	Pitfalls in variant annotation for hereditary cancer diagnostics: The example of IlluminaÂ® VariantStudioÂ®. Genomics, 2021, 113, 748-754.	2.9	0

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91	Identifying and testing for hereditary susceptibility to breast/ovarian cancer in Serbia: Where are we now?. Archive of Oncology, 2006, 14, 131-135.	0.2	0