

Katri Pylkäs

List of Publications by Year in descending order

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Version: 2024-02-01

122
papers

13,151
citations

50566

48
h-index

29333

108
g-index

126
all docs

126
docs citations

126
times ranked

18589
citing authors

#	ARTICLE	IF	CITATIONS
1	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017, 551, 92-94.	13.7	1,099
2	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013, 45, 353-361.	9.4	960
3	Breast-Cancer Risk in Families with Mutations in <i>PALB2</i> . <i>New England Journal of Medicine</i> , 2014, 371, 497-506.	13.9	745
4	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019, 104, 21-34.	2.6	711
5	Associations of Breast Cancer Risk Factors With Tumor Subtypes: A Pooled Analysis From the Breast Cancer Association Consortium Studies. <i>Journal of the National Cancer Institute</i> , 2011, 103, 250-263.	3.0	596
6	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014, 514, 92-97.	13.7	548
7	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. <i>Nature Genetics</i> , 2015, 47, 373-380.	9.4	513
8	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , 2013, 45, 371-384.	9.4	493
9	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. <i>Journal of the National Cancer Institute</i> , 2015, 107, .	3.0	428
10	A recurrent mutation in <i>PALB2</i> in Finnish cancer families. <i>Nature</i> , 2007, 446, 316-319.	13.7	402
11	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013, 45, 392-398.	9.4	374
12	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. <i>Nature Genetics</i> , 2015, 47, 1294-1303.	9.4	357
13	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, 885-892.	9.4	309
14	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017, 49, 1767-1778.	9.4	289
15	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. <i>Nature Genetics</i> , 2011, 43, 1210-1214.	9.4	279
16	Cancer Risks Associated With Germline <i>PALB2</i> Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020, 38, 674-685.	0.8	270
17	Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012, 44, 312-318.	9.4	256
18	Functional Variants at the 11q13 Risk Locus for Breast Cancer Regulate Cyclin D1 Expression through Long-Range Enhancers. <i>American Journal of Human Genetics</i> , 2013, 92, 489-503.	2.6	201

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19	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. <i>Nature Genetics</i> , 2018, 50, 968-978.	9.4	184
20	RAD50 and NBS1 are breast cancer susceptibility genes associated with genomic instability. <i>Carcinogenesis</i> , 2005, 27, 1593-1599.	1.3	179
21	<i>PALB2</i> , <i>CHEK2</i> and <i>ATM</i> rare variants and cancer risk: data from COGS. <i>Journal of Medical Genetics</i> , 2016, 53, 800-811.	1.5	174
22	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. <i>Nature Genetics</i> , 2016, 48, 374-386.	9.4	125
23	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020, 52, 56-73.	9.4	120
24	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. <i>PLoS Medicine</i> , 2016, 13, e1002105.	3.9	118
25	Common Breast Cancer Susceptibility Loci Are Associated with Triple-Negative Breast Cancer. <i>Cancer Research</i> , 2011, 71, 6240-6249.	0.4	109
26	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. <i>Nature Medicine</i> , 2021, 27, 1012-1024.	15.2	109
27	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014, 5, 4999.	5.8	105
28	19p13.1 Is a Triple-Negative-Specific Breast Cancer Susceptibility Locus. <i>Cancer Research</i> , 2012, 72, 1795-1803.	0.4	100
29	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015, 107, djv219.	3.0	99
30	Fine-Scale Mapping of the FGFR2 Breast Cancer Risk Locus: Putative Functional Variants Differentially Bind FOXA1 and E2F1. <i>American Journal of Human Genetics</i> , 2013, 93, 1046-1060.	2.6	98
31	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. <i>Breast Cancer Research</i> , 2014, 16, 3419.	2.2	97
32	No evidence that protein truncating variants in <i>BRIP1</i> are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016, 53, 298-309.	1.5	94
33	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016, 7, 11375.	5.8	93
34	Penetrance Analysis of the <i>PALB2</i> c.1592delT Founder Mutation. <i>Clinical Cancer Research</i> , 2008, 14, 4667-4671.	3.2	90
35	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. <i>International Journal of Epidemiology</i> , 2019, 48, 795-806.	0.9	81
36	The role of genetic breast cancer susceptibility variants as prognostic factors. <i>Human Molecular Genetics</i> , 2012, 21, 3926-3939.	1.4	80

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37	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016, 7, 12675.	5.8	78
38	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016, 108, djv315.	3.0	77
39	Fine-Scale Mapping of the 5q11.2 Breast Cancer Locus Reveals at Least Three Independent Risk Variants Regulating MAP3K1. <i>American Journal of Human Genetics</i> , 2015, 96, 5-20.	2.6	76
40	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017, 77, 2789-2799.	0.4	75
41	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2011, 20, 4693-4706.	1.4	71
42	Heterozygous mutations in PALB2 cause DNA replication and damage response defects. <i>Nature Communications</i> , 2013, 4, 2578.	5.8	60
43	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. <i>American Journal of Human Genetics</i> , 2016, 99, 903-911.	2.6	59
44	Machine learning identifies interacting genetic variants contributing to breast cancer risk: A case study in Finnish cases and controls. <i>Scientific Reports</i> , 2018, 8, 13149.	1.6	58
45	Identification of Novel Genetic Markers of Breast Cancer Survival. <i>Journal of the National Cancer Institute</i> , 2015, 107, .	3.0	56
46	Further evidence for the contribution of the RAD51C gene in hereditary breast and ovarian cancer susceptibility. <i>Breast Cancer Research and Treatment</i> , 2011, 130, 1003-1010.	1.1	54
47	Breast Cancer-Associated Abraxas Mutation Disrupts Nuclear Localization and DNA Damage Response Functions. <i>Science Translational Medicine</i> , 2012, 4, 122ra23.	5.8	54
48	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2014, 23, 6096-6111.	1.4	53
49	Genome-wide association study of germline variants and breast cancer-specific mortality. <i>British Journal of Cancer</i> , 2019, 120, 647-657.	2.9	52
50	Comparison of 6q25 Breast Cancer Hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). <i>PLoS ONE</i> , 2012, 7, e42380.	1.1	51
51	MicroRNA Related Polymorphisms and Breast Cancer Risk. <i>PLoS ONE</i> , 2014, 9, e109973.	1.1	49
52	Body mass index and breast cancer survival: a Mendelian randomization analysis. <i>International Journal of Epidemiology</i> , 2017, 46, 1814-1822.	0.9	45
53	Genetic predisposition to ductal carcinoma in situ of the breast. <i>Breast Cancer Research</i> , 2016, 18, 22.	2.2	43
54	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. <i>Breast Cancer Research</i> , 2017, 19, 119.	2.2	43

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55	Association of common ATM polymorphism with bilateral breast cancer. <i>International Journal of Cancer</i> , 2005, 116, 69-72.	2.3	42
56	Analysis of large deletions in BRCA1, BRCA2 and PALB2 genes in Finnish breast and ovarian cancer families. <i>BMC Cancer</i> , 2008, 8, 146.	1.1	42
57	Inactivation of Palb2 gene leads to mesoderm differentiation defect and early embryonic lethality in mice. <i>Human Molecular Genetics</i> , 2010, 19, 3021-3029.	1.4	41
58	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. <i>Circulation</i> , 2018, 137, 2716-2726.	1.6	41
59	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. <i>Human Molecular Genetics</i> , 2015, 24, 2966-2984.	1.4	40
60	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. <i>PLoS Genetics</i> , 2014, 10, e1004285.	1.5	39
61	Patient survival and tumor characteristics associated with CHEK2:p.1157T findings from the Breast Cancer Association Consortium. <i>Breast Cancer Research</i> , 2016, 18, 98.	2.2	39
62	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. <i>American Journal of Human Genetics</i> , 2020, 107, 837-848.	2.6	39
63	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. <i>Human Molecular Genetics</i> , 2015, 24, 285-298.	1.4	38
64	KEAP1 Genetic Polymorphisms Associate with Breast Cancer Risk and Survival Outcomes. <i>Clinical Cancer Research</i> , 2015, 21, 1591-1601.	3.2	37
65	11q13 is a susceptibility locus for hormone receptor positive breast cancer. <i>Human Mutation</i> , 2012, 33, 1123-1132.	1.1	35
66	An intergenic risk locus containing an enhancer deletion in 2q35 modulates breast cancer risk by deregulating IGFBP5 expression. <i>Human Molecular Genetics</i> , 2016, 25, 3863-3876.	1.4	33
67	A large-scale assessment of two-way SNP interactions in breast cancer susceptibility using 46 450 cases and 42 461 controls from the breast cancer association consortium. <i>Human Molecular Genetics</i> , 2014, 23, 1934-1946.	1.4	32
68	Transcriptome-wide association study of breast cancer risk by estrogen receptor status. <i>Genetic Epidemiology</i> , 2020, 44, 442-468.	0.6	32
69	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. <i>Oncotarget</i> , 2016, 7, 80140-80163.	0.8	31
70	Rare Copy Number Variants Observed in Hereditary Breast Cancer Cases Disrupt Genes in Estrogen Signaling and TP53 Tumor Suppression Network. <i>PLoS Genetics</i> , 2012, 8, e1002734.	1.5	28
71	Confirmation of 5p12 As a Susceptibility Locus for Progesterone-Receptor Positive, Lower Grade Breast Cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011, 20, 2222-2231.	1.1	27
72	Common germline polymorphisms associated with breast cancer-specific survival. <i>Breast Cancer Research</i> , 2015, 17, 58.	2.2	26

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73	FANCM mutation c.5791C>T is a risk factor for triple-negative breast cancer in the Finnish population. <i>Breast Cancer Research and Treatment</i> , 2017, 166, 217-226.	1.1	26
74	Screening for large genomic rearrangements in the FANCA gene reveals extensive deletion in a Finnish breast cancer family. <i>Cancer Letters</i> , 2011, 302, 113-118.	3.2	22
75	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. <i>PLoS Genetics</i> , 2016, 12, e1005816.	1.5	22
76	Evaluation of the role of Finnish ataxia-telangiectasia mutations in hereditary predisposition to breast cancer. <i>Carcinogenesis</i> , 2006, 28, 1040-1045.	1.3	21
77	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. <i>Cancer Causes and Control</i> , 2016, 27, 679-693.	0.8	21
78	SNP-SNP interaction analysis of NF- κ B signaling pathway on breast cancer survival. <i>Oncotarget</i> , 2015, 6, 37979-37994.	0.8	20
79	Case-control analysis of truncating mutations in DNA damage response genes connects TEX15 and FANCD2 with hereditary breast cancer susceptibility. <i>Scientific Reports</i> , 2017, 7, 681.	1.6	20
80	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). <i>Scientific Reports</i> , 2016, 6, 32512.	1.6	19
81	The <i>BRCA2</i> c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018, 39, 729-741.	1.1	19
82	Genome-wide search for breast cancer linkage in large Icelandic non-BRCA1/2 families. <i>Breast Cancer Research</i> , 2010, 12, R50.	2.2	18
83	Recurrent CYP2C19 deletion allele is associated with triple-negative breast cancer. <i>BMC Cancer</i> , 2014, 14, 902.	1.1	18
84	9q31.2-rs865686 as a Susceptibility Locus for Estrogen Receptor-Positive Breast Cancer: Evidence from the Breast Cancer Association Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 1783-1791.	1.1	17
85	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. <i>Clinical Genetics</i> , 2015, 88, 68-73.	1.0	17
86	2q36.3 is associated with prognosis for oestrogen receptor-negative breast cancer patients treated with chemotherapy. <i>Nature Communications</i> , 2014, 5, 4051.	5.8	16
87	Novel variants and phenotypes widen the phenotypic spectrum of GABRG2-related disorders. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 69, 99-104.	0.9	16
88	Screening for RAD51 and BRCA2 BRC repeat mutations in breast and ovarian cancer families. <i>Cancer Letters</i> , 2006, 236, 142-147.	3.2	15
89	Germline alterations in the CLSPN gene in breast cancer families. <i>Cancer Letters</i> , 2008, 261, 93-97.	3.2	15
90	Rare missense mutations in <i>RECQL</i> and <i>POLG</i> associate with inherited predisposition to breast cancer. <i>International Journal of Cancer</i> , 2018, 142, 2286-2292.	2.3	15

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91	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. <i>Breast Cancer Research</i> , 2022, 24, 2.	2.2	15
92	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. <i>Breast Cancer Research</i> , 2014, 16, R51.	2.2	14
93	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. <i>Carcinogenesis</i> , 2015, 36, 256-271.	1.3	14
94	Screening for large genomic rearrangements of the BRIP1 and CHK1 genes in Finnish breast cancer families. <i>Familial Cancer</i> , 2010, 9, 537-540.	0.9	13
95	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. <i>International Journal of Cancer</i> , 2016, 139, 2760-2770.	2.3	13
96	Germline alterations in the 53BP1 gene in breast and ovarian cancer families. <i>Cancer Letters</i> , 2007, 245, 337-340.	3.2	12
97	Mutation screening of the MERIT40 gene encoding a novel BRCA1 and RAP80 interacting protein in breast cancer families. <i>Breast Cancer Research and Treatment</i> , 2010, 120, 165-168.	1.1	12
98	Evaluation of the need for routine clinical testing of PALB2 c.1592delT mutation in BRCA negative Northern Finnish breast cancer families. <i>BMC Medical Genetics</i> , 2013, 14, 82.	2.1	12
99	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. <i>Human Molecular Genetics</i> , 2014, 23, 6034-6046.	1.4	12
100	Exome sequencing identifies a recurrent variant in SERPINA3 associating with hereditary susceptibility to breast cancer. <i>European Journal of Cancer</i> , 2021, 143, 46-51.	1.3	11
101	<i>PHIP</i> - a novel candidate breast cancer susceptibility locus on 6q14.1. <i>Oncotarget</i> , 2017, 8, 102769-102782.	0.8	9
102	Mutation analysis of the AATF gene in breast cancer families. <i>BMC Cancer</i> , 2009, 9, 457.	1.1	8
103	The UGT1A6_19_GC genotype is a breast cancer risk factor. <i>Frontiers in Genetics</i> , 2013, 4, 104.	1.1	8
104	Assessment of Targeted and Non-Targeted Responses in Cells Deficient in ATM Function following Exposure to Low and High Dose X-Rays. <i>PLoS ONE</i> , 2014, 9, e93211.	1.1	7
105	Haplotypes of the I157T CHEK2 germline mutation in ethnically diverse populations. <i>Familial Cancer</i> , 2009, 8, 473-478.	0.9	6
106	Mutation analysis of the gene encoding the PALB2-binding protein MRG15 in BRCA1/2-negative breast cancer families. <i>Journal of Human Genetics</i> , 2010, 55, 842-843.	1.1	6
107	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. <i>American Journal of Human Genetics</i> , 2021, 108, 1190-1203.	2.6	6
108	Rare germline copy number variants (CNVs) and breast cancer risk. <i>Communications Biology</i> , 2022, 5, 65.	2.0	6

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109	A novel variant in SMG9 causes intellectual disability, confirming a role for nonsense-mediated decay components in neurocognitive development. <i>European Journal of Human Genetics</i> , 2022, 30, 619-627.	1.4	6
110	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. <i>International Journal of Cancer</i> , 2019, 145, 2070-2081.	2.3	5
111	Evaluating the role of <i>NTHL1</i> p.Q90* allele in inherited breast cancer predisposition. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1493.	0.6	5
112	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. <i>British Journal of Cancer</i> , 2021, 124, 842-854.	2.9	5
113	Mutation screening of the RNF8, UBC13 and MMS2 genes in Northern Finnish breast cancer families. <i>BMC Medical Genetics</i> , 2011, 12, 98.	2.1	4
114	BRCA1 mislocalization leads to aberrant DNA damage response in heterozygous ABRAXAS1 mutation carrier cells. <i>Human Molecular Genetics</i> , 2019, 28, 4148-4160.	1.4	4
115	Genetic Variants Associated With Sudden Cardiac Death in Victims With Single Vessel Coronary Artery Disease and Left Ventricular Hypertrophy With or Without Fibrosis. <i>Frontiers in Cardiovascular Medicine</i> , 2021, 8, 755062.	1.1	3
116	rs2735383, located at a microRNA binding site in the 3'UTR of NBS1, is not associated with breast cancer risk. <i>Scientific Reports</i> , 2016, 6, 36874.	1.6	2
117	Evaluating the role of MLH3 p.Ser1188Ter variant in inherited breast cancer predisposition. <i>Genetics in Medicine</i> , 2020, 22, 663-664.	1.1	2
118	Novel Rare SORL1 Variants in Early-Onset Dementia. <i>Journal of Alzheimer's Disease</i> , 2021, 82, 761-770.	1.2	2
119	Breast-Cancer Risk in Families With Mutations in PALB2. <i>Obstetrical and Gynecological Survey</i> , 2014, 69, 659-660.	0.2	1
120	Genetic contributions to the expression of acquired causes of cardiac hypertrophy in non-ischemic sudden cardiac death victims. <i>Scientific Reports</i> , 2021, 11, 11171.	1.6	1
121	Truncating TINF2 p.Tyr312Ter variant and inherited breast cancer susceptibility. <i>Familial Cancer</i> , 2022, , .	0.9	1
122	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. <i>Obstetrical and Gynecological Survey</i> , 2015, 70, 758-762.	0.2	0