

Katri Pylks

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

122
papers

9,795
citations

44
h-index

98
g-index

126
ext. papers

11,792
ext. citations

11.4
avg, IF

3.91
L-index

#	Paper	IF	Citations
122	Rare germline copy number variants (CNVs) and breast cancer risk.. <i>Communications Biology</i> , 2022 , 5, 65	6.7	0
121	Common variants in breast cancer risk loci predispose to distinct tumor subtypes.. <i>Breast Cancer Research</i> , 2022 , 24, 2	8.3	3
120	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 ,	5.3	1
119	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	5.4	0
118	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	4.9	
117	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. <i>Nature Medicine</i> , 2021 , 27, 1012-1024	50.5	16
116	Novel Rare SORL1 Variants in Early-Onset Dementia. <i>Journal of Alzheimer's Disease</i> , 2021 , 82, 761-770	4.3	1
115	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	11	1
114	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	8.7	2
113	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	7.5	1
112	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. <i>Genetic Epidemiology</i> , 2020 , 44, 442-468	2.6	9
111	Evaluating the role of MLH3 p.Ser1188Ter variant in inherited breast cancer predisposition. <i>Genetics in Medicine</i> , 2020 , 22, 663-664	8.1	
110	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. <i>Nature Genetics</i> , 2020 , 52, 56-73	36.3	56
109	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. <i>Journal of Clinical Oncology</i> , 2020 , 38, 674-685	2.2	133
108	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. <i>American Journal of Human Genetics</i> , 2020 , 107, 837-848	11	12
107	Evaluating the role of NTHL1 p.Q90* allele in inherited breast cancer predisposition. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1493	2.3	3
106	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	3.2	11

105	Genome-wide association study of germline variants and breast cancer-specific mortality. <i>British Journal of Cancer</i> , 2019 , 120, 647-657	8.7	28
104	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. <i>International Journal of Cancer</i> , 2019 , 145, 2070-2081	7.5	3
103	BRCA1 mislocalization leads to aberrant DNA damage response in heterozygous ABRAXAS1 mutation carrier cells. <i>Human Molecular Genetics</i> , 2019 , 28, 4148-4160	5.6	2
102	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. <i>American Journal of Human Genetics</i> , 2019 , 104, 21-34	11	363
101	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	7.8	52
100	The BRCA2 c.68-7T>A variant is not pathogenic: A model for clinical calibration of spliceogenicity. <i>Human Mutation</i> , 2018 , 39, 729-741	4.7	16
99	Rare missense mutations in RECQL and POLG associate with inherited predisposition to breast cancer. <i>International Journal of Cancer</i> , 2018 , 142, 2286-2292	7.5	11
98	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	36.3	101
97	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. <i>Circulation</i> , 2018 , 137, 2716-2726	16.7	22
96	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	4.9	31
95	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. <i>Cancer Research</i> , 2017 , 77, 2789-2799	10.1	49
94	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	4.9	10
93	Association analysis identifies 65 new breast cancer risk loci. <i>Nature</i> , 2017 , 551, 92-94	50.4	643
92	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. <i>Nature Genetics</i> , 2017 , 49, 1767-1778	36.3	186
91	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. <i>Breast Cancer Research</i> , 2017 , 19, 119	8.3	26
90	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	4.4	17
89	Body mass index and breast cancer survival: a Mendelian randomization analysis. <i>International Journal of Epidemiology</i> , 2017 , 46, 1814-1822	7.8	27
88	- a novel candidate breast cancer susceptibility locus on 6q14.1. <i>Oncotarget</i> , 2017 , 8, 102769-102782	3.3	3

87	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	5.6	24
86	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. <i>International Journal of Cancer</i> , 2016 , 139, 2760-2770	7.5	10
85	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	4.9	2
84	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , 2016 , 7, 11375	17.4	64
83	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. <i>Nature Communications</i> , 2016 , 7, 12675	17.4	53
82	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	4.9	16
81	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. <i>Journal of Medical Genetics</i> , 2016 , 53, 298-309	5.8	83
80	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
79	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. <i>Journal of the National Cancer Institute</i> , 2016 , 108,	9.7	65
78	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	3.3	21
77	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	11.6	80
76	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. <i>PLoS Genetics</i> , 2016 , 12, e1005816	6	17
75	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. <i>Journal of Medical Genetics</i> , 2016 , 53, 800-811	5.8	121
74	Patient survival and tumor characteristics associated with CHEK2:p.I157T - findings from the Breast Cancer Association Consortium. <i>Breast Cancer Research</i> , 2016 , 18, 98	8.3	26
73	Genetic predisposition to ductal carcinoma in situ of the breast. <i>Breast Cancer Research</i> , 2016 , 18, 22	8.3	31
72	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, 679-93	2.8	15
71	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	11	43
70	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, 256-71	4.6	12

69	KEAP1 Genetic Polymorphisms Associate with Breast Cancer Risk and Survival Outcomes. <i>Clinical Cancer Research</i> , 2015 , 21, 1591-601	12.9	29
68	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. <i>Nature Genetics</i> , 2015 , 47, 373-80	36.3	406
67	Identification of novel genetic markers of breast cancer survival. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	38
66	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	36.3	226
65	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	74
64	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, 285-98	5.6	35
63	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. <i>Clinical Genetics</i> , 2015 , 88, 68-73	4	13
62	Common germline polymorphisms associated with breast cancer-specific survival. <i>Breast Cancer Research</i> , 2015 , 17, 58	8.3	24
61	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 ²⁻⁴		
60	SNP-SNP interaction analysis of NF- κ B signaling pathway on breast cancer survival. <i>Oncotarget</i> , 2015 , 6, 37979-94	3.3	19
59	Prediction of breast cancer risk based on profiling with common genetic variants. <i>Journal of the National Cancer Institute</i> , 2015 , 107,	9.7	324
58	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. <i>Human Molecular Genetics</i> , 2015 , 24, 2966-84	5.6	36
57	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	11	59
56	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, 1934-46	5.6	28
55	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , 2014 , 514, 92-97	50.4	401
54	Breast-cancer risk in families with mutations in PALB2. <i>New England Journal of Medicine</i> , 2014 , 371, 497-506	59.6	576
53	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. <i>Nature Communications</i> , 2014 , 4, 4999	17.4	87
52	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. <i>Human Molecular Genetics</i> , 2014 , 23, 6034-46	5.6	11

51	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	8.3	12
50	MicroRNA related polymorphisms and breast cancer risk. <i>PLoS ONE</i> , 2014 , 9, e109973	3.7	37
49	Genetic predisposition to in situ and invasive lobular carcinoma of the breast. <i>PLoS Genetics</i> , 2014 , 10, e1004285	6	38
48	2q36.3 is associated with prognosis for oestrogen receptor-negative breast cancer patients treated with chemotherapy. <i>Nature Communications</i> , 2014 , 5, 4051	17.4	13
47	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. <i>Human Molecular Genetics</i> , 2014 , 23, 6096-111	5.6	48
46	Recurrent CYP2C19 deletion allele is associated with triple-negative breast cancer. <i>BMC Cancer</i> , 2014 , 14, 902	4.8	12
45	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	8.3	82
44	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	3.7	7
43	Heterozygous mutations in PALB2 cause DNA replication and damage response defects. <i>Nature Communications</i> , 2013 , 4, 2578	17.4	56
42	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, 1046-60	11	80
41	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, 371-84, 384e1-2	36.3	422
40	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	9
39	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	11	167
38	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. <i>Nature Genetics</i> , 2013 , 45, 392-8, 398e1-2	36.3	327
37	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. <i>Nature Genetics</i> , 2013 , 45, 353-61, 361e1-2	36.3	813
36	The UGT1A6_19_GG genotype is a breast cancer risk factor. <i>Frontiers in Genetics</i> , 2013 , 4, 104	4.5	7
35	Genome-wide association analysis identifies three new breast cancer susceptibility loci. <i>Nature Genetics</i> , 2012 , 44, 312-8	36.3	237
34	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, 1783-91	4	17

33	11q13 is a susceptibility locus for hormone receptor positive breast cancer. <i>Human Mutation</i> , 2012 , 33, 1123-32	4.7	33
32	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	6	25
31	Breast cancer-associated Abraxas mutation disrupts nuclear localization and DNA damage response functions. <i>Science Translational Medicine</i> , 2012 , 4, 122ra23	17.5	43
30	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. <i>Cancer Research</i> , 2012 , 72, 1795-803	10.1	93
29	The role of genetic breast cancer susceptibility variants as prognostic factors. <i>Human Molecular Genetics</i> , 2012 , 21, 3926-39	5.6	75
28	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	3.7	49
27	Screening for large genomic rearrangements in the FANCA gene reveals extensive deletion in a Finnish breast cancer family. <i>Cancer Letters</i> , 2011 , 302, 113-8	9.9	17
26	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, 1003-10	4.4	46
25	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	3
24	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2011 , 20, 2222-31	4	27
23	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, 250-63	9.7	513
22	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. <i>Nature Genetics</i> , 2011 , 43, 1210-4	36.3	253
21	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, 4693-706	5.6	66
20	Common breast cancer susceptibility loci are associated with triple-negative breast cancer. <i>Cancer Research</i> , 2011 , 71, 6240-9	10.1	100
19	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
18	Inactivation of Palb2 gene leads to mesoderm differentiation defect and early embryonic lethality in mice. <i>Human Molecular Genetics</i> , 2010 , 19, 3021-9	5.6	31
17	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, 842-3	4.3	4
16	Genome-wide search for breast cancer linkage in large Icelandic non-BRCA1/2 families. <i>Breast Cancer Research</i> , 2010 , 12, R50	8.3	15

15	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, 165-8	4.4	12
14	Screening for large genomic rearrangements of the BRIP1 and CHK1 genes in Finnish breast cancer families. <i>Familial Cancer</i> , 2010 , 9, 537-40	3	12
13	Mutation analysis of the AATF gene in breast cancer families. <i>BMC Cancer</i> , 2009 , 9, 457	4.8	5
12	Haplotypes of the I157T CHEK2 germline mutation in ethnically diverse populations. <i>Familial Cancer</i> , 2009 , 8, 473-8	3	5
11	Analysis of large deletions in BRCA1, BRCA2 and PALB2 genes in Finnish breast and ovarian cancer families. <i>BMC Cancer</i> , 2008 , 8, 146	4.8	37
10	Germline alterations in the CLSPN gene in breast cancer families. <i>Cancer Letters</i> , 2008 , 261, 93-7	9.9	10
9	Penetrance analysis of the PALB2 c.1592delT founder mutation. <i>Clinical Cancer Research</i> , 2008 , 14, 4667-71	17.9	84
8	A recurrent mutation in PALB2 in Finnish cancer families. <i>Nature</i> , 2007 , 446, 316-9	50.4	349
7	Germline alterations in the 53BP1 gene in breast and ovarian cancer families. <i>Cancer Letters</i> , 2007 , 245, 337-40	9.9	11
6	Evaluation of the role of Finnish ataxia-telangiectasia mutations in hereditary predisposition to breast cancer. <i>Carcinogenesis</i> , 2007 , 28, 1040-5	4.6	16
5	RAD50 and NBS1 are breast cancer susceptibility genes associated with genomic instability. <i>Carcinogenesis</i> , 2006 , 27, 1593-9	4.6	145
4	Screening for RAD51 and BRCA2 BRC repeat mutations in breast and ovarian cancer families. <i>Cancer Letters</i> , 2006 , 236, 142-7	9.9	13
3	Association of common ATM polymorphism with bilateral breast cancer. <i>International Journal of Cancer</i> , 2005 , 116, 69-72	7.5	34
2	Common variants in breast cancer risk loci predispose to distinct tumor subtypes		1
1	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2