Katri Pylks

List of Publications by Citations

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	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
121	Association analysis identifies 65 new breast cancer risk loci. *Nature*, **2017**, 551, 92-94	50.4	643
120	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
119	Associations of breast cancer risk factors with tumor subtypes: a pooled analysis from the Breast Cancer Association Consortium studies. *Journal of the National Cancer Institute*, **2011**, 103, 250-63	9.7	513
118	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
117	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
116	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. *Nature*, **2014**, 514, 92-97	50.4	401
115	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
114	A recurrent mutation in PALB2 in Finnish cancer families. *Nature*, **2007**, 446, 316-9	50.4	349
113	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
112	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
111	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. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
110	A common variant at the TERT-CLPTM1L locus is associated with estrogen receptor-negative breast cancer. *Nature Genetics*, **2011**, 43, 1210-4	36.3	253
109	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
108	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
107	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
106	Functional variants at the 11q13 risk locus for breast cancer regulate cyclin D1 expression through long-range enhancers. *American Journal of Human Genetics*, **2013**, 92, 489-503	11	167

105	RAD50 and NBS1 are breast cancer susceptibility genes associated with genomic instability. *Carcinogenesis*, **2006**, 27, 1593-9	4.6	145
104	Cancer Risks Associated With Germline Pathogenic Variants: An International Study of 524 Families. *Journal of Clinical Oncology*, **2020**, 38, 674-685	2.2	133
103	PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS. *Journal of Medical Genetics*, **2016**, 53, 800-811	5.8	121
102	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. *Nature Genetics*, **2018**, 50, 968-978	36.3	101
101	Common breast cancer susceptibility loci are associated with triple-negative breast cancer. *Cancer Research*, **2011**, 71, 6240-9	10.1	100
100	Breast cancer risk variants at 6q25 display different phenotype associations and regulate ESR1, RMND1 and CCDC170. *Nature Genetics*, **2016**, 48, 374-86	36.3	93
99	19p13.1 is a triple-negative-specific breast cancer susceptibility locus. *Cancer Research*, **2012**, 72, 1795-803	10.1	93
98	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. *Nature Communications*, **2014**, 4, 4999	17.4	87
97	Penetrance analysis of the PALB2 c.1592delT founder mutation. *Clinical Cancer Research*, **2008**, 14, 4667-71	12.9	84
96	No evidence that protein truncating variants in BRIP1 are associated with breast cancer risk: implications for gene panel testing. *Journal of Medical Genetics*, **2016**, 53, 298-309	5.8	83
95	Refined histopathological predictors of BRCA1 and BRCA2 mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. *Breast Cancer Research* , **2014**, 16, 3419	8.3	82
94	Fine-scale mapping of the FGFR2 breast cancer risk locus: putative functional variants differentially bind FOXA1 and E2F1. *American Journal of Human Genetics*, **2013**, 93, 1046-60	11	80
93	Genetically Predicted Body Mass Index and Breast Cancer Risk: Mendelian Randomization Analyses of Data from 145,000 Women of European Descent. *PLoS Medicine*, **2016**, 13, e1002105	11.6	80
92	The role of genetic breast cancer susceptibility variants as prognostic factors. *Human Molecular Genetics*, **2012**, 21, 3926-39	5.6	75
91	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	74
90	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. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
89	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. *Journal of the National Cancer Institute*, **2016**, 108,	9.7	65
88	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64

87	Fine-scale mapping of the 5q11.2 breast cancer locus reveals at least three independent risk variants regulating MAP3K1. *American Journal of Human Genetics*, **2015**, 96, 5-20	11	59
86	Heterozygous mutations in PALB2 cause DNA replication and damage response defects. *Nature Communications*, **2013**, 4, 2578	17.4	56
85	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
84	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breast-ovarian cancer susceptibility locus. *Nature Communications*, **2016**, 7, 12675	17.4	53
83	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2019**, 48, 795-806	7.8	52
82	Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. *Cancer Research*, **2017**, 77, 2789-2799	10.1	49
81	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
80	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2014**, 23, 6096-111	5.6	48
79	Further evidence for the contribution of the RAD51C gene in hereditary breast and ovarian cancer susceptibility. *Breast Cancer Research and Treatment*, **2011**, 130, 1003-10	4.4	46
78	Breast cancer-associated Abraxas mutation disrupts nuclear localization and DNA damage response functions. *Science Translational Medicine*, **2012**, 4, 122ra23	17.5	43
77	Evidence that the 5p12 Variant rs10941679 Confers Susceptibility to Estrogen-Receptor-Positive Breast Cancer through FGF10 and MRPS30 Regulation. *American Journal of Human Genetics*, **2016**, 99, 903-911	11	43
76	Identification of novel genetic markers of breast cancer survival. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	38
75	Genetic predisposition to in situ and invasive lobular carcinoma of the breast. *PLoS Genetics*, **2014**, 10, e1004285	6	38
74	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
73	Analysis of large deletions in BRCA1, BRCA2 and PALB2 genes in Finnish breast and ovarian cancer families. *BMC Cancer*, **2008**, 8, 146	4.8	37
72	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. *Human Molecular Genetics*, **2015**, 24, 2966-84	5.6	36
71	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
70	Association of common ATM polymorphism with bilateral breast cancer. *International Journal of Cancer*, **2005**, 116, 69-72	7.5	34

69	11q13 is a susceptibility locus for hormone receptor positive breast cancer. *Human Mutation*, **2012**, 33, 1123-32	4.7	33
68	Inactivation of Palb2 gene leads to mesoderm differentiation defect and early embryonic lethality in mice. *Human Molecular Genetics*, **2010**, 19, 3021-9	5.6	31
67	Genetic predisposition to ductal carcinoma in situ of the breast. *Breast Cancer Research*, **2016**, 18, 22	8.3	31
66	Machine learning identifies interacting genetic variants contributing to breast cancer risk: A case study in Finnish cases and controls. *Scientific Reports*, **2018**, 8, 13149	4.9	31
65	KEAP1 Genetic Polymorphisms Associate with Breast Cancer Risk and Survival Outcomes. *Clinical Cancer Research*, **2015**, 21, 1591-601	12.9	29
64	Genome-wide association study of germline variants and breast cancer-specific mortality. *British Journal of Cancer*, **2019**, 120, 647-657	8.7	28
63	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. *Human Molecular Genetics*, **2014**, 23, 1934-46	5.6	28
62	Body mass index and breast cancer survival: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2017**, 46, 1814-1822	7.8	27
61	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
60	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. *Breast Cancer Research*, **2017**, 19, 119	8.3	26
59	Patient survival and tumor characteristics associated with CHEK2:p.I157T - findings from the Breast Cancer Association Consortium. *Breast Cancer Research*, **2016**, 18, 98	8.3	26
58	Rare copy number variants observed in hereditary breast cancer cases disrupt genes in estrogen signaling and TP53 tumor suppression network. *PLoS Genetics*, **2012**, 8, e1002734	6	25
57	An intergenic risk locus containing an enhancer deletion in 2q35 modulates breast cancer risk by deregulating IGFBP5 expression. *Human Molecular Genetics*, **2016**, 25, 3863-3876	5.6	24
56	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
55	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. *Circulation*, **2018**, 137, 2716-2726	16.7	22
54	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. *Oncotarget*, **2016**, 7, 80140-80163	3.3	21
53	SNP-SNP interaction analysis of NF-B signaling pathway on breast cancer survival. *Oncotarget*, **2015**, 6, 37979-94	3.3	19
52	FANCM mutation c.5791C>T is a risk factor for triple-negative breast cancer in the Finnish population. *Breast Cancer Research and Treatment*, **2017**, 166, 217-226	4.4	17

51	9q31.2-rs865686 as a susceptibility locus for estrogen receptor-positive breast cancer: evidence from the Breast Cancer Association Consortium. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1783-91	4	17
50	Screening for large genomic rearrangements in the FANCA gene reveals extensive deletion in a Finnish breast cancer family. *Cancer Letters*, **2011**, 302, 113-8	9.9	17
49	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. *PLoS Genetics*, **2016**, 12, e1005816	6	17
48	The BRCA2 c.68-7T□□A variant is not pathogenic: A model for clinical calibration of spliceogenicity. *Human Mutation*, **2018**, 39, 729-741	4.7	16
47	Fine scale mapping of the 17q22 breast cancer locus using dense SNPs, genotyped within the Collaborative Oncological Gene-Environment Study (COGs). *Scientific Reports*, **2016**, 6, 32512	4.9	16
46	Evaluation of the role of Finnish ataxia-telangiectasia mutations in hereditary predisposition to breast cancer. *Carcinogenesis*, **2007**, 28, 1040-5	4.6	16
45	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. *Nature Medicine*, **2021**, 27, 1012-1024	50.5	16
44	Genome-wide search for breast cancer linkage in large Icelandic non-BRCA1/2 families. *Breast Cancer Research*, **2010**, 12, R50	8.3	15
43	Association of genetic susceptibility variants for type 2 diabetes with breast cancer risk in women of European ancestry. *Cancer Causes and Control*, **2016**, 27, 679-93	2.8	15
42	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. *Clinical Genetics*, **2015**, 88, 68-73	4	13
41	2q36.3 is associated with prognosis for oestrogen receptor-negative breast cancer patients treated with chemotherapy. *Nature Communications*, **2014**, 5, 4051	17.4	13
40	Screening for RAD51 and BRCA2 BRC repeat mutations in breast and ovarian cancer families. *Cancer Letters*, **2006**, 236, 142-7	9.9	13
39	Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer. *Carcinogenesis*, **2015**, 36, 256-71	4.6	12
38	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. *Breast Cancer Research*, **2014**, 16, R51	8.3	12
37	Recurrent CYP2C19 deletion allele is associated with triple-negative breast cancer. *BMC Cancer*, **2014**, 14, 902	4.8	12
36	Mutation screening of the MERIT40 gene encoding a novel BRCA1 and RAP80 interacting protein in breast cancer families. *Breast Cancer Research and Treatment*, **2010**, 120, 165-8	4.4	12
35	Screening for large genomic rearrangements of the BRIP1 and CHK1 genes in Finnish breast cancer families. *Familial Cancer*, **2010**, 9, 537-40	3	12
34	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 837-848	11	12

33	Novel variants and phenotypes widen the phenotypic spectrum of GABRG2-related disorders. *Seizure: the Journal of the British Epilepsy Association*, **2019**, 69, 99-104	3.2	11
32	Rare missense mutations in RECQL and POLG associate with inherited predisposition to breast cancer. *International Journal of Cancer*, **2018**, 142, 2286-2292	7.5	11
31	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. *Human Molecular Genetics*, **2014**, 23, 6034-46	5.6	11
30	Germline alterations in the 53BP1 gene in breast and ovarian cancer families. *Cancer Letters*, **2007**, 245, 337-40	9.9	11
29	Case-control analysis of truncating mutations in DNA damage response genes connects TEX15 and FANCD2 with hereditary breast cancer susceptibility. *Scientific Reports*, **2017**, 7, 681	4.9	10
28	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. *International Journal of Cancer*, **2016**, 139, 2760-2770	7.5	10
27	Germline alterations in the CLSPN gene in breast cancer families. *Cancer Letters*, **2008**, 261, 93-7	9.9	10
26	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
25	Evaluation of the need for routine clinical testing of PALB2 c.1592delT mutation in BRCA negative Northern Finnish breast cancer families. *BMC Medical Genetics*, **2013**, 14, 82	2.1	9
24	The UGT1A6_19_GG genotype is a breast cancer risk factor. *Frontiers in Genetics*, **2013**, 4, 104	4.5	7
23	Assessment of targeted and non-targeted responses in cells deficient in ATM function following exposure to low and high dose X-rays. *PLoS ONE*, **2014**, 9, e93211	3.7	7
22	Mutation analysis of the AATF gene in breast cancer families. *BMC Cancer*, **2009**, 9, 457	4.8	5
21	Haplotypes of the I157T CHEK2 germline mutation in ethnically diverse populations. *Familial Cancer* , **2009**, 8, 473-8	3	5
20	Mutation analysis of the gene encoding the PALB2-binding protein MRG15 in BRCA1/2-negative breast cancer families. *Journal of Human Genetics*, **2010**, 55, 842-3	4.3	4
19	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. *International Journal of Cancer*, **2019**, 145, 2070-2081	7.5	3
18	Mutation screening of the RNF8, UBC13 and MMS2 genes in Northern Finnish breast cancer families. *BMC Medical Genetics*, **2011**, 12, 98	2.1	3
17	Common variants in breast cancer risk loci predispose to distinct tumor subtypes.. *Breast Cancer Research*, **2022**, 24, 2	8.3	3
16	- a novel candidate breast cancer susceptibility locus on 6q14.1. *Oncotarget*, **2017**, 8, 102769-102782	3.3	3

15	Evaluating the role of NTHL1 p.Q90* allele in inherited breast cancer predisposition. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1493	2.3	3
14	rs2735383, located at a microRNA binding site in the 3′UTR of NBS1, is not associated with breast cancer risk. *Scientific Reports*, **2016**, 6, 36874	4.9	2
13	BRCA1 mislocalization leads to aberrant DNA damage response in heterozygous ABRAXAS1 mutation carrier cells. *Human Molecular Genetics*, **2019**, 28, 4148-4160	5.6	2
12	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
11	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. *British Journal of Cancer*, **2021**, 124, 842-854	8.7	2
10	A novel variant in SMG9 causes intellectual disability, confirming a role for nonsense-mediated decay components in neurocognitive development.. *European Journal of Human Genetics*, **2022**,	5.3	1
9	Common variants in breast cancer risk loci predispose to distinct tumor subtypes		1
8	Novel Rare SORL1 Variants in Early-Onset Dementia. *Journal of Alzheimer's Disease*, **2021**, 82, 761-770	4.3	1
7	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. *American Journal of Human Genetics*, **2021**, 108, 1190-1203	11	1
6	Exome sequencing identifies a recurrent variant in SERPINA3 associating with hereditary susceptibility to breast cancer. *European Journal of Cancer*, **2021**, 143, 46-51	7.5	1
5	Rare germline copy number variants (CNVs) and breast cancer risk.. *Communications Biology*, **2022**, 5, 65	6.7	0
4	Genetic Variants Associated With Sudden Cardiac Death in Victims With Single Vessel Coronary Artery Disease and Left Ventricular Hypertrophy With or Without Fibrosis.. *Frontiers in Cardiovascular Medicine*, **2021**, 8, 755062	5.4	0
3	Large-Scale Genomic Analyses Link Reproductive Aging to Hypothalamic Signaling, Breast Cancer Susceptibility, and BRCA1-Mediated DNA Repair. *Obstetrical and Gynecological Survey*, **2015**, 70, 758-762	2.4	
2	Evaluating the role of MLH3 p.Ser1188Ter variant in inherited breast cancer predisposition. *Genetics in Medicine*, **2020**, 22, 663-664	8.1	
1	Genetic contributions to the expression of acquired causes of cardiac hypertrophy in non-ischemic sudden cardiac death victims. *Scientific Reports*, **2021**, 11, 11171	4.9	