Katri PylkÃ¤s

List of Publications by Year in descending order

122 papers	13,151 citations	44069 48 h-index	25787 108 g-index
126 all docs	126 docs citations	126 times ranked	17018 citing authors

#	Article	IF	Citations
1	Rare germline copy number variants (CNVs) and breast cancer risk. Communications Biology, 2022, 5, 65.	4.4	6
2	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer Research, 2022, 24, 2.	5.0	15
3	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, 30, 619-627.	2.8	6
4	Truncating TINF2 p.Tyr312Ter variant and inherited breast cancer susceptibility. Familial Cancer, 2022, , .	1.9	1
5	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.	6.4	5
6	Exome sequencing identifies a recurrent variant in SERPINA3 associating with hereditary susceptibility to breast cancer. European Journal of Cancer, 2021, 143, 46-51.	2.8	11
7	Genetic contributions to the expression of acquired causes of cardiac hypertrophy in non-ischemic sudden cardiac death victims. Scientific Reports, 2021, 11, 11171.	3.3	1
8	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	30.7	109
9	Novel Rare SORL1 Variants in Early-Onset Dementia. Journal of Alzheimer's Disease, 2021, 82, 761-770.	2.6	2
10	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.	6.2	6
11	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.	2.4	3
12	Evaluating the role of MLH3 p.Ser1188Ter variant in inherited breast cancer predisposition. Genetics in Medicine, 2020, 22, 663-664.	2.4	2
13	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
14	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
15	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
16	Evaluating the role of NTHL1 p.Q90* allele in inherited breast cancer predisposition. Molecular Genetics & Genomic Medicine, 2020, 8, e1493.	1.2	5
17	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
18	BRCA1 mislocalization leads to aberrant DNA damage response in heterozygous ABRAXAS1 mutation carrier cells. Human Molecular Genetics, 2019, 28, 4148-4160.	2.9	4

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19	Novel variants and phenotypes widen the phenotypic spectrum of GABRG2-related disorders. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 99-104.	2.0	16
20	Genome-wide association study of germline variants and breast cancer-specific mortality. British Journal of Cancer, 2019, 120, 647-657.	6.4	52
21	Tumor suppressor MCPH1 regulates gene expression profiles related to malignant conversion and chromosomal assembly. International Journal of Cancer, 2019, 145, 2070-2081.	5.1	5
22	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
23	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	1.9	81
24	The BRCA2 c.68-7TÂ>ÂA variant is not pathogenic: A model for clinical calibration of spliceogenicity. Human Mutation, 2018, 39, 729-741.	2.5	19
25	Rare missense mutations in RECQL and POLG associate with inherited predisposition to breast cancer. International Journal of Cancer, 2018, 142, 2286-2292.	5.1	15
26	Machine learning identifies interacting genetic variants contributing to breast cancer risk: A case study in Finnish cases and controls. Scientific Reports, 2018, 8, 13149.	3.3	58
27	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	21.4	184
28	Primary Myocardial Fibrosis as an Alternative Phenotype Pathway of Inherited Cardiac Structural Disorders. Circulation, 2018, 137, 2716-2726.	1.6	41
29	BRCA2 Hypomorphic Missense Variants Confer Moderate Risks of Breast Cancer. Cancer Research, 2017, 77, 2789-2799.	0.9	75
30	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.	3.3	20
31	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
32	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
33	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.	2.5	26
34	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	1.9	45
35	Reproductive profiles and risk of breast cancer subtypes: a multi-center case-only study. Breast Cancer Research, 2017, 19, 119.	5.0	43
36	PHIP- a novel candidate breast cancer susceptibility locus on 6q14.1. Oncotarget, 2017, 8, 102769-102782.	1.8	9

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37	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.	1.8	31
38	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.	8.4	118
39	Targeted Next-Generation Sequencing Identifies a Recurrent Mutation in MCPH1 Associating with Hereditary Breast Cancer Susceptibility. PLoS Genetics, 2016, 12, e1005816.	3.5	22
40	PALB2,CHEK2andATMrare variants and cancer risk: data from COGS. Journal of Medical Genetics, 2016, 53, 800-811.	3.2	174
41	Patient survival and tumor characteristics associated with CHEK2:p.I157T â€“ findings from the Breast Cancer Association Consortium. Breast Cancer Research, 2016, 18, 98.	5.0	39
42	Genetic predisposition to ductal carcinoma in situ of the breast. Breast Cancer Research, 2016, 18, 22.	5.0	43
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, 679-693.	1.8	21
44	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.	6.2	59
45	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.	2.9	33
46	FANCM c.5101C>T mutation associates with breast cancer survival and treatment outcome. International Journal of Cancer, 2016, 139, 2760-2770.	5.1	13
47	rs2735383, located at a microRNA binding site in the 3â€™UTR of NBS1, is not associated with breast cancer risk. Scientific Reports, 2016, 6, 36874.	3.3	2
48	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
49	Functional mechanisms underlying pleiotropic risk alleles at the 19p13.1 breastâ€“ovarian cancer susceptibility locus. Nature Communications, 2016, 7, 12675.	12.8	78
50	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.	3.3	19
51	No evidence that protein truncating variants inBRIP1are associated with breast cancer risk: implications for gene panel testing. Journal of Medical Genetics, 2016, 53, 298-309.	3.2	94
52	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
53	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
54	Common germline polymorphisms associated with breast cancer-specific survival. Breast Cancer Research, 2015, 17, 58.	5.0	26

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55	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.	0.4	0
56	SNP-SNP interaction analysis of NF-ÎºB signaling pathway on breast cancer survival. Oncotarget, 2015, 6, 37979-37994.	1.8	20
57	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	6.3	428
58	Fine-mapping identifies two additional breast cancer susceptibility loci at 9q31.2. Human Molecular Genetics, 2015, 24, 2966-2984.	2.9	40
59	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.	6.2	76
60	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, 256-271.	2.8	14
61	KEAP1 Genetic Polymorphisms Associate with Breast Cancer Risk and Survival Outcomes. Clinical Cancer Research, 2015, 21, 1591-1601.	7.0	37
62	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	21.4	513
63	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	6.3	56
64	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	21.4	357
65	Height and Breast Cancer Risk: Evidence From Prospective Studies and Mendelian Randomization. Journal of the National Cancer Institute, 2015, 107, djv219.	6.3	99
66	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. Human Molecular Genetics, 2015, 24, 285-298.	2.9	38
67	Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer. Clinical Genetics, 2015, 88, 68-73.	2.0	17
68	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	2.5	49
69	Genetic Predisposition to In Situ and Invasive Lobular Carcinoma of the Breast. PLoS Genetics, 2014, 10, e1004285.	3.5	39
70	2q36.3 is associated with prognosis for oestrogen receptor-negative breast cancer patients treated with chemotherapy. Nature Communications, 2014, 5, 4051.	12.8	16
71	Common non-synonymous SNPs associated with breast cancer susceptibility: findings from the Breast Cancer Association Consortium. Human Molecular Genetics, 2014, 23, 6096-6111.	2.9	53
72	Recurrent CYP2C19 deletion allele is associated with triple-negative breast cancer. BMC Cancer, 2014, 14, 902.	2.6	18

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73	Refined histopathological predictors of BRCA1 and BRCA2mutation status: a large-scale analysis of breast cancer characteristics from the BCAC, CIMBA, and ENIGMA consortia. Breast Cancer Research, 2014, 16, 3419.	5.0	97
74	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.4	1
75	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, 1934-1946.	2.9	32
76	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. Nature, 2014, 514, 92-97.	27.8	548
77	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
78	Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation. Nature Communications, 2014, 5, 4999.	12.8	105
79	Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade. Human Molecular Genetics, 2014, 23, 6034-6046.	2.9	12
80	Genetic variation at CYP3A is associated with age at menarche and breast cancer risk: a case-control study. Breast Cancer Research, 2014, 16, R51.	5.0	14
81	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.	2.5	7
82	Heterozygous mutations in PALB2 cause DNA replication and damage response defects. Nature Communications, 2013, 4, 2578.	12.8	60
83	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, 1046-1060.	6.2	98
84	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
85	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	12
86	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.	6.2	201
87	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	21.4	374
88	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	21.4	960
89	The UGT1A6_19_GG genotype is a breast cancer risk factor. Frontiers in Genetics, 2013, 4, 104.	2.3	8
90	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.	3.5	28

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91	Breast Cancerâ€“Associated Abraxas Mutation Disrupts Nuclear Localization and DNA Damage Response Functions. Science Translational Medicine, 2012, 4, 122ra23.	12.4	54
92	19p13.1 Is a Triple-Negativeâ€“Specific Breast Cancer Susceptibility Locus. Cancer Research, 2012, 72, 1795-1803.	0.9	100
93	The role of genetic breast cancer susceptibility variants as prognostic factors. Human Molecular Genetics, 2012, 21, 3926-3939.	2.9	80
94	Genome-wide association analysis identifies three new breast cancer susceptibility loci. Nature Genetics, 2012, 44, 312-318.	21.4	256
95	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, 1783-1791.	2.5	17
96	11q13 is a susceptibility locus for hormone receptor positive breast cancer. Human Mutation, 2012, 33, 1123-1132.	2.5	35
97	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.	2.5	51
98	Confirmation of 5p12 As a Susceptibility Locus for Progesterone-Receptorâ€“Positive, Lower Grade Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 2222-2231.	2.5	27
99	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, 250-263.	6.3	596
100	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
101	Screening for large genomic rearrangements in the FANCA gene reveals extensive deletion in a Finnish breast cancer family. Cancer Letters, 2011, 302, 113-118.	7.2	22
102	Further evidence for the contribution of the RAD51C gene in hereditary breast and ovarian cancer susceptibility. Breast Cancer Research and Treatment, 2011, 130, 1003-1010.	2.5	54
103	Mutation screening of the RNF8, UBC13 and MMS2 genes in Northern Finnish breast cancer families. BMC Medical Genetics, 2011, 12, 98.	2.1	4
104	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 Consortiumâ€. Human Molecular Genetics, 2011, 20, 4693-4706.	2.9	71
105	Common Breast Cancer Susceptibility Loci Are Associated with Triple-Negative Breast Cancer. Cancer Research, 2011, 71, 6240-6249.	0.9	109
106	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, 165-168.	2.5	12
107	Screening for large genomic rearrangements of the BRIP1 and CHK1 genes in Finnish breast cancer families. Familial Cancer, 2010, 9, 537-540.	1.9	13
108	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	21.4	309

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109	Inactivation of Palb2 gene leads to mesoderm differentiation defect and early embryonic lethality in mice. Human Molecular Genetics, 2010, 19, 3021-3029.	2.9	41
110	Mutation analysis of the gene encoding the PALB2-binding protein MRG15 in BRCA1/2-negative breast cancer families. Journal of Human Genetics, 2010, 55, 842-843.	2.3	6
111	Genome-wide search for breast cancer linkage in large Icelandic non-BRCA1/2 families. Breast Cancer Research, 2010, 12, R50.	5.0	18
112	Mutation analysis of the AATF gene in breast cancer families. BMC Cancer, 2009, 9, 457.	2.6	8
113	Haplotypes of the I157T CHEK2 germline mutation in ethnically diverse populations. Familial Cancer, 2009, 8, 473-478.	1.9	6
114	Analysis of large deletions in BRCA1, BRCA2 and PALB2 genes in Finnish breast and ovarian cancer families. BMC Cancer, 2008, 8, 146.	2.6	42
115	Germline alterations in the CLSPN gene in breast cancer families. Cancer Letters, 2008, 261, 93-97.	7.2	15
116	Penetrance Analysis of the PALB2 c.1592delT Founder Mutation. Clinical Cancer Research, 2008, 14, 4667-4671.	7.0	90
117	Germline alterations in the 53BP1 gene in breast and ovarian cancer families. Cancer Letters, 2007, 245, 337-340.	7.2	12
118	A recurrent mutation in PALB2 in Finnish cancer families. Nature, 2007, 446, 316-319.	27.8	402
119	Evaluation of the role of Finnish ataxia-telangiectasia mutations in hereditary predisposition to breast cancer. Carcinogenesis, 2006, 28, 1040-1045.	2.8	21
120	Screening for RAD51 and BRCA2 BRC repeat mutations in breast and ovarian cancer families. Cancer Letters, 2006, 236, 142-147.	7.2	15
121	Association of commonATM polymorphism with bilateral breast cancer. International Journal of Cancer, 2005, 116, 69-72.	5.1	42
122	RAD50 and NBS1 are breast cancer susceptibility genes associated with genomic instability. Carcinogenesis, 2005, 27, 1593-1599.	2.8	179