Inge Skilde Pedersen

List of Publications by Citations

76 papers	2,637 citations	26 h-index	50 g-index
81 ext. papers	3,439 ext. citations	6.5 avg, IF	3.62 L-index

#	Paper	IF	Citations
76	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
75	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	4.7	211
74	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209
73	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. *Nature Genetics*, **2017**, 49, 680-691	36.3	190
72	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
71	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. *Human Mutation*, **2018**, 39, 593-620	4.7	138
70	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. *Journal of Clinical Oncology*, **2017**, 35, 2240-2250	2.2	101
69	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
68	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. *Breast Cancer Research*, **2016**, 18, 15	8.3	58
67	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. *Nature Genetics*, **2020**, 52, 56-73	36.3	56
66	Comparison of mRNA splicing assay protocols across multiple laboratories: recommendations for best practice in standardized clinical testing. *Clinical Chemistry*, **2014**, 60, 341-52	5.5	53
65	Serological and molecular evidence of Rickettsia helvetica in Denmark. *Scandinavian Journal of Infectious Diseases*, **2004**, 36, 559-63		53
64	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. *Human Mutation*, **2019**, 40, 1557-1578	4.7	52
63	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. *Breast Cancer Research and Treatment*, **2012**, 132, 1009-23	4.4	51
62	Biallelic expression of the IGF2 gene in human breast disease. *Human Molecular Genetics*, **1996**, 5, 1123-7	5.6	48
61	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. *Nature Communications*, **2019**, 10, 1741	17.4	47
60	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	8.3	46

59	Hypermethylated DNA, a circulating biomarker for colorectal cancer detection. *PLoS ONE*, **2017**, 12, e0180809	3.7	45
58	Cell-free DNA promoter hypermethylation in plasma as a diagnostic marker for pancreatic adenocarcinoma. *Clinical Epigenetics*, **2016**, 8, 117	7.7	45
57	Hypermethylated DNA as a biomarker for colorectal cancer: a systematic review. *Colorectal Disease*, **2016**, 18, 549-61	2.1	41
56	BRCA1 and BRCA2 mutations in Danish families with hereditary breast and/or ovarian cancer. *Acta Oncologica*, **2008**, 47, 772-7	3.2	40
55	Promoter switch: a novel mechanism causing biallelic PEG1/MEST expression in invasive breast cancer. *Human Molecular Genetics*, **2002**, 11, 1449-53	5.6	39
54	The c. 5096G>A p.Arg1699Gln (R1699Q) intermediate risk variant: breast and ovarian cancer risk estimation and recommendations for clinical management from the ENIGMA consortium. *Journal of Medical Genetics*, **2018**, 55, 15-20	5.8	36
53	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1653-1666	8.1	34
52	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33
51	COMPLEXO: identifying the missing heritability of breast cancer via next generation collaboration. *Breast Cancer Research*, **2013**, 15, 402	8.3	30
50	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
49	Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). *JAMA Oncology*, **2020**, 6, 1218-1230	13.4	25
48	Mutations in CARD15 and smoking confer susceptibility to Crohn's disease in the Danish population. *Scandinavian Journal of Gastroenterology*, **2007**, 42, 1445-51	2.4	24
47	ECEL1 mutation causes fetal arthrogryposis multiplex congenita. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 731-43	2.5	23
46	Cell-free DNA promoter hypermethylation in plasma as a predictive marker for survival of patients with pancreatic adenocarcinoma. *Oncotarget*, **2017**, 8, 93942-93956	3.3	23
45	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. *Cancer Research*, **2020**, 80, 624-638	10.1	22
44	The MAS proto-oncogene is imprinted in human breast tissue. *Genomics*, **1997**, 46, 509-12	4.3	21
43	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
42	Whole-exome sequencing identifies a GREB1L variant in a three-generation family with Müllerian and renal agenesis: a novel candidate gene in Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. A case report. *Human Reproduction*, **2019**, 34, 1838-1846	5.7	17

41	Promoter hypermethylation in plasma-derived cell-free DNA as a prognostic marker for pancreatic adenocarcinoma staging. *International Journal of Cancer*, **2017**, 141, 2489-2497	7.5	17
40	High recovery of cell-free methylated DNA based on a rapid bisulfite-treatment protocol. *BMC Molecular Biology*, **2012**, 13, 12	4.5	16
39	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	8.3	16
38	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. *Gynecologic Oncology*, **2016**, 141, 386-401	4.9	15
37	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. *Breast Cancer Research and Treatment*, **2017**, 161, 117-134	4.4	15
36	Full in-frame exon 3 skipping of confers high risk of breast and/or ovarian cancer. *Oncotarget*, **2018**, 9, 17334-17348	3.3	13
35	Clinical and molecular characterization of BRCA-associated breast cancer: results from the DBCG. *Acta Oncologica*, **2018**, 57, 95-101	3.2	12
34	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and Breast/Ovarian) Cancer Susceptibility Genes: An International Survey by the Evidence-Based Network for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. *JCO Precision Oncology*, **2018**, 2,	3.6	12
33	The prognostic efficacy of cell-free DNA hypermethylation in colorectal cancer. *Oncotarget*, **2018**, 9, 7010-7022	3.3	11
32	Genetic variants of glutathione S-transferases mu, theta, and pi display no susceptibility to inflammatory bowel disease in the Danish population. *Scandinavian Journal of Gastroenterology*, **2010**, 45, 1068-75	2.4	10
31	Transcriptome-wide association study of breast cancer risk by estrogen-receptor status. *Genetic Epidemiology*, **2020**, 44, 442-468	2.6	9
30	BRCA1/BRCA2 founder mutations and cancer risks: impact in the western Danish population. *Familial Cancer*, **2016**, 15, 507-12	3	8
29	Cancer Risks Associated With and Pathogenic Variants.. *Journal of Clinical Oncology*, **2022**, JCO2102112	2.2	7
28	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. *PLoS ONE*, **2016**, 11, e0158801	3.7	7
27	Development of a Precision Medicine Workflow in Hematological Cancers, Aalborg University Hospital, Denmark. *Cancers*, **2020**, 12,	6.6	6
26	Clinical outcomes of female breast cancer according to BRCA mutation status. *Cancer Epidemiology*, **2017**, 49, 128-137	2.8	5
25	Mitochondrial Disease Caused by a Novel Homozygous Mutation (Gly106del) in the SCO1 Gene. *Neonatology*, **2019**, 116, 290-294	4	5
24	De novo Mutation Identified with Whole-Exome Sequencing in a Girl with Say-Barber/Biesecker/Young-Simpson Syndrome. *Molecular Syndromology*, **2017**, 8, 24-29	1.5	4

23	The PHF6 Mutation c.1A>G; pM1V Causes B□jeson-Forsman-Lehmann Syndrome in a Family with Four Affected Young Boys. *Molecular Syndromology*, **2015**, 6, 181-6	1.5	4
22	Common polymorphisms in the microsomal epoxide hydrolase and N-acetyltransferase 2 genes in association with inflammatory bowel disease in the Danish population. *European Journal of Gastroenterology and Hepatology*, **2011**, 23, 269-74	2.2	4
21	A BRCA2 mutation incorrectly mapped in the original BRCA2 reference sequence, is a common West Danish founder mutation disrupting mRNA splicing. *Breast Cancer Research and Treatment*, **2011**, 128, 179-85	4.4	4
20	A case-only study to identify genetic modifiers of breast cancer risk for BRCA1/BRCA2 mutation carriers. *Nature Communications*, **2021**, 12, 1078	17.4	4
19	DNA methylation changes in genes involved in inflammation and depression in fibromyalgia: a pilot study. *Scandinavian Journal of Pain*, **2021**, 21, 372-383	1.9	3
18	Association of germline variation with the survival of women with pathogenic variants and breast cancer. *Npj Breast Cancer*, **2020**, 6, 44	7.8	3
17	Cell-based non-invasive prenatal testing for monogenic disorders: confirmation of unaffected fetuses following preimplantation genetic testing. *Journal of Assisted Reproduction and Genetics*, **2021**, 38, 1959-1970	3.4	3
16	Mutational landscape of immune surveillance genes in diffuse large B-cell lymphoma. *Expert Review of Hematology*, **2020**, 13, 655-668	2.8	3
15	Differential effect of surgical manipulation on gene expression in normal breast tissue and breast tumor tissue. *Molecular Medicine*, **2018**, 24, 57	6.2	3
14	Breast and Prostate Cancer Risks for Male BRCA1 and BRCA2 Pathogenic Variant Carriers Using Polygenic Risk Scores. *Journal of the National Cancer Institute*, **2021**,	9.7	3
13	Epigenetic Regulation of Ferroportin in Primary Cultures of the Rat Blood-Brain Barrier. *Molecular Neurobiology*, **2020**, 57, 3526-3539	6.2	2
12	A systematic review on concurrent aneuploidy screening and preimplantation genetic testing for hereditary disorders: What is the prevalence of aneuploidy and is there a clinical effect from aneuploidy screening?. *Acta Obstetricia Et Gynecologica Scandinavica*, **2020**, 99, 696-706	3.8	2
11	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants.. *Genetics in Medicine*, **2021**,	8.1	2
10	DNA methylation biomarkers in peripheral blood of patients with head and neck squamous cell carcinomas. A systematic review. *PLoS ONE*, **2020**, 15, e0244101	3.7	2
9	Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes		2
8	A Danish national effort of BRCA1/2 variant classification. *Acta Oncol□gica*, **2018**, 57, 159-162	3.2	2
7	DNA Methylation Changes in Fibromyalgia Suggest the Role of the Immune-Inflammatory Response and Central Sensitization. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
6	Polygenic Risk Modelling for Prediction of Epithelial Ovarian Cancer Risk		1

5	Dysfibrinogenemia-Potential Impact of Genotype on Thrombosis or Bleeding. *Seminars in Thrombosis and Hemostasis*, **2021**,	5.3	1
4	Use of genetic testing for hypolactasia trait in the North Denmark Region. *Scandinavian Journal of Gastroenterology*, **2020**, 55, 1012-1018	2.4	0
3	Prevalence and type distribution of human papillomavirus infections in Danish patients diagnosed with vulvar squamous cell tumors and precursors. *Gynecologic Oncology Reports*, **2021**, 37, 100828	1.3	0
2	The mutational profile of immune surveillance genes in diagnostic and refractory/relapsed DLBCLs. *BMC Cancer*, **2021**, 21, 829	4.8	
1	A family-based study to identify genetic biomarkers of fibromyalgia: consideration of patientsQ subgroups. *Clinical and Experimental Rheumatology*, **2021**, 39 Suppl 130, 144-152	2.2	