## Anne-Marie Gerdes

## List of Publications by Citations

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#	Paper	IF	Citations
136	Risks of Breast, Ovarian, and Contralateral Breast Cancer for BRCA1 and BRCA2 Mutation Carriers.  JAMA - Journal of the American Medical Association, 2017, 317, 2402-2416	27.4	1140
135	Long-term effect of aspirin on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. <i>Lancet, The</i> , <b>2011</b> , 378, 2081-7	40	715
134	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. <i>Nature Genetics</i> , <b>2013</b> , 45, 371-84, 384e1-2	36.3	422
133	Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 134-47	4	411
132	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. <i>JAMA - Journal of the American Medical Association</i> , <b>2015</b> , 313, 1347-61	27.4	286
131	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> , <b>2010</b> , 42, 885-92	36.3	276
130	Diagnostic criteria for constitutional mismatch repair deficiency syndrome: suggestions of the European consortium 'care for CMMRD' (C4CMMRD). <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 355-65	5.8	274
129	Common breast cancer-predisposition alleles are associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 937-48	11	218
128	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003212	6	209
127	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. <i>Nature Genetics</i> , <b>2017</b> , 49, 680-691	36.3	190
126	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer.  Nature Genetics, 2017, 49, 1767-1778	36.3	186
125	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. <i>Nature Genetics</i> , <b>2015</b> , 47, 164-71	36.3	177
124	Deep sequencing of uveal melanoma identifies a recurrent mutation in PLCB4. <i>Oncotarget</i> , <b>2016</b> , 7, 462	24 <del>5</del> .31	168
123	Functional Significance and Clinical Phenotype of Nontruncating Mismatch Repair Variants of MLH1. <i>Gastroenterology</i> , <b>2005</b> , 129, 537-549	13.3	158
122	Common breast cancer susceptibility alleles and the risk of breast cancer for BRCA1 and BRCA2 mutation carriers: implications for risk prediction. <i>Cancer Research</i> , <b>2010</b> , 70, 9742-54	10.1	147
121	Mutational spectrum in a worldwide study of 29,700 families with BRCA1 or BRCA2 mutations. <i>Human Mutation</i> , <b>2018</b> , 39, 593-620	4.7	138
120	Multiple self-healing squamous epithelioma is caused by a disease-specific spectrum of mutations in TGFBR1. <i>Nature Genetics</i> , <b>2011</b> , 43, 365-9	36.3	119

119	Tamoxifen and risk of contralateral breast cancer for BRCA1 and BRCA2 mutation carriers. <i>Journal of Clinical Oncology</i> , <b>2013</b> , 31, 3091-9	2.2	118
118	Oral contraceptives and breast cancer risk in the international BRCA1/2 carrier cohort study: a report from EMBRACE, GENEPSO, GEO-HEBON, and the IBCCS Collaborating Group. <i>Journal of Clinical Oncology</i> , <b>2007</b> , 25, 3831-6	2.2	116
117	Reproductive and hormonal factors, and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers: results from the International BRCA1/2 Carrier Cohort Study. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2009</b> , 18, 601-10	4	110
116	Nonsense mutations in the shelterin complex genes ACD and TERF2IP in familial melanoma. <i>Journal of the National Cancer Institute</i> , <b>2015</b> , 107,	9.7	102
115	A cryptic BAP1 splice mutation in a family with uveal and cutaneous melanoma, and paraganglioma. <i>Pigment Cell and Melanoma Research</i> , <b>2012</b> , 25, 815-8	4.5	96
114	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 4442-56	5.6	91
113	Cancer prevention with aspirin in hereditary colorectal cancer (Lynch syndrome), 10-year follow-up and registry-based 20-year data in the CAPP2 study: a double-blind, randomised, placebo-controlled trial. <i>Lancet, The</i> , <b>2020</b> , 395, 1855-1863	40	88
112	Chromosome abnormalities in benign hyperproliferative disorders of epithelial and stromal breast tissue. <i>International Journal of Cancer</i> , <b>1995</b> , 60, 49-53	7.5	85
111	Functional significance and clinical phenotype of nontruncating mismatch repair variants of MLH1. <i>Gastroenterology</i> , <b>2005</b> , 129, 537-49	13.3	79
110	Classifications within molecular subtypes enables identification of BRCA1/BRCA2 mutation carriers by RNA tumor profiling. <i>PLoS ONE</i> , <b>2013</b> , 8, e64268	3.7	71
109	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2012</b> , 14, R33	8.3	70
108	Long-term effect of resistant starch on cancer risk in carriers of hereditary colorectal cancer: an analysis from the CAPP2 randomised controlled trial. <i>Lancet Oncology, The</i> , <b>2012</b> , 13, 1242-9	21.7	70
107	Chromosome abnormalities in bilateral breast carcinomas. Cytogenetic evaluation of the clonal origin of multiple primary tumors. <i>Cancer</i> , <b>1995</b> , 76, 250-8	6.4	69
106	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. <i>Nature Communications</i> , <b>2016</b> , 7, 11375	17.4	64
105	Diagnosis of Constitutional Mismatch Repair-Deficiency Syndrome Based on Microsatellite Instability and Lymphocyte Tolerance to Methylating Agents. <i>Gastroenterology</i> , <b>2015</b> , 149, 1017-29.e3	13.3	63
104	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110	8.3	62
103	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 3304-21	5.6	62
102	Male breast cancer in BRCA1 and BRCA2 mutation carriers: pathology data from the Consortium of Investigators of Modifiers of BRCA1/2. <i>Breast Cancer Research</i> , <b>2016</b> , 18, 15	8.3	58

101	Pathogenicity of MSH2 missense mutations is typically associated with impaired repair capability of the mutated protein. <i>Gastroenterology</i> , <b>2006</b> , 131, 1408-17	13.3	54
100	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. <i>Human Mutation</i> , <b>2019</b> , 40, 1557-1578	4.7	52
99	Hereditary breast cancer: clinical, pathological and molecular characteristics. <i>Breast Cancer: Basic and Clinical Research</i> , <b>2014</b> , 8, 145-55	2.2	50
98	A BAP1 mutation in a Danish family predisposes to uveal melanoma and other cancers. <i>PLoS ONE</i> , <b>2013</b> , 8, e72144	3.7	48
97	Functional characterization of BRCA1 gene variants by mini-gene splicing assay. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 1362-8	5.3	46
96	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2014</b> , 16, 3416	8.3	46
95	Common variants at the 19p13.1 and ZNF365 loci are associated with ER subtypes of breast cancer and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2012</b> , 21, 645-57	4	44
94	Organochlorines, p53 mutations in relation to breast cancer risk and survival. A Danish cohort-nested case-controls study. <i>Breast Cancer Research and Treatment</i> , <b>2002</b> , 71, 59-65	4.4	44
93	HNPCC mutation MLH1 P648S makes the functional protein unstable, and homozygosity predisposes to mild neurofibromatosis type 1. <i>Genes Chromosomes and Cancer</i> , <b>2004</b> , 40, 261-5	5	43
92	Low frequency of large genomic rearrangements of BRCA1 and BRCA2 in western Denmark. <i>Cancer Genetics and Cytogenetics</i> , <b>2006</b> , 168, 168-71		41
91	BRCA1 and BRCA2 mutations in Danish families with hereditary breast and/or ovarian cancer. <i>Acta Oncoligica</i> , <b>2008</b> , 47, 772-7	3.2	40
90	POLE mutations in families predisposed to cutaneous melanoma. Familial Cancer, 2015, 14, 621-8	3	38
89	Current perspectives on recommendations for BRCA genetic testing in ovarian cancer patients. <i>European Journal of Cancer</i> , <b>2016</b> , 69, 127-134	7.5	37
88	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Journal of the National Cancer Institute</i> , <b>2011</b> , 103, 105-16	9.7	37
87	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. <i>Journal of Medical Genetics</i> , <b>2018</b> , 55, 15-20	5.8	36
86	Toward mechanistic models for genotype-phenotype correlations in phenylketonuria using protein stability calculations. <i>Human Mutation</i> , <b>2019</b> , 40, 444-457	4.7	36
85	Germline TERT promoter mutations are rare in familial melanoma. Familial Cancer, 2016, 15, 139-44	3	34
84	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1653-1666	8.1	34

83	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004256	6	33	
82	Incidence, prevalence, etiology, and prognosis of first-time chronic pancreatitis in young patients: a nationwide cohort study. <i>Digestive Diseases and Sciences</i> , <b>2010</b> , 55, 2988-98	4	33	
81	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. <i>Human Mutation</i> , <b>2012</b> , 33, 690-702	4.7	31	
80	Is screening for pancreatic cancer in high-risk groups cost-effective? - Experience from a Danish national screening program. <i>Pancreatology</i> , <b>2016</b> , 16, 584-92	3.8	31	
79	Major contribution from recurrent alterations and MSH6 mutations in the Danish Lynch syndrome population. <i>Familial Cancer</i> , <b>2009</b> , 8, 75-83	3	30	
78	Genetic, epidemiological, and clinical aspects of hereditary pancreatitis: a population-based cohort study in Denmark. <i>American Journal of Gastroenterology</i> , <b>2010</b> , 105, 1876-83	0.7	28	
77	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. <i>PLoS ONE</i> , <b>2015</b> , 10, e0120020	3.7	26	
76	Cytogenetic abnormalities in an in situ ductal carcinoma and five prophylactically removed breasts from members of a family with hereditary breast cancer. <i>Breast Cancer Research and Treatment</i> , <b>1996</b> , 38, 177-82	4.4	26	
75	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). <i>JAMA Oncology</i> , <b>2020</b> , 6, 1218-1230	13.4	25	
74	Prophylactic total gastrectomy in hereditary diffuse gastric cancer: identification of two novel CDH1 gene mutations-a clinical observational study. <i>Familial Cancer</i> , <b>2014</b> , 13, 231-42	3	23	
73	Risk-reducing salpingo-oophorectomy, natural menopause, and breast cancer risk: an international prospective cohort of BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2020</b> , 22, 8	8.3	22	
7 <sup>2</sup>	Association of Genomic Domains in and with Prostate Cancer Risk and Aggressiveness. <i>Cancer Research</i> , <b>2020</b> , 80, 624-638	10.1	22	
71	An international survey of surveillance schemes for unaffected BRCA1 and BRCA2 mutation carriers. <i>Breast Cancer Research and Treatment</i> , <b>2016</b> , 157, 319-327	4.4	22	
70	Identification of six pathogenic RAD51C mutations via mutational screening of 1228 Danish individuals with increased risk of hereditary breast and/or ovarian cancer. <i>Breast Cancer Research and Treatment</i> , <b>2016</b> , 155, 215-22	4.4	21	
69	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. <i>Breast Cancer Research and Treatment</i> , <b>2011</b> , 127, 671-9	4.4	21	
68	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 4732-47	5.6	21	
67	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2015</b> , 24, 308-16	4	20	
66	A nonsynonymous polymorphism in IRS1 modifies risk of developing breast and ovarian cancers in BRCA1 and ovarian cancer in BRCA2 mutation carriers. <i>Cancer Epidemiology Biomarkers and Prevention</i> <b>2012</b> 21 1362-70	4	20	

65	Multiple self-healing squamous epithelioma in different ethnic groups: more than a founder mutation disorder?. <i>Journal of Investigative Dermatology</i> , <b>2007</b> , 127, 2336-44	4.3	20
64	Blocking protein quality control to counter hereditary cancers. <i>Genes Chromosomes and Cancer</i> , <b>2017</b> , 56, 823-831	5	18
63	Computational and cellular studies reveal structural destabilization and degradation of MLH1 variants in Lynch syndrome. <i>ELife</i> , <b>2019</b> , 8,	8.9	17
62	Functional examination of MLH1, MSH2, and MSH6 intronic mutations identified in Danish colorectal cancer patients. <i>BMC Medical Genetics</i> , <b>2013</b> , 14, 103	2.1	16
61	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. <i>Breast Cancer Research</i> , <b>2015</b> , 17, 61	8.3	16
60	Identification of a Danish breast/ovarian cancer family double heterozygote for BRCA1 and BRCA2 mutations. <i>Familial Cancer</i> , <b>2010</b> , 9, 283-7	3	16
59	Nationwide germline whole genome sequencing of 198 consecutive pediatric cancer patients reveals a high incidence of cancer prone syndromes. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1009231	6	16
58	Molecular characterization of melanoma cases in Denmark suspected of genetic predisposition. <i>PLoS ONE</i> , <b>2015</b> , 10, e0122662	3.7	16
57	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. <i>Gynecologic Oncology</i> , <b>2016</b> , 141, 386-401	4.9	15
56	Is DBCG abreast of new developments?. Acta Oncolgica, 2018, 57, 1-2	3.2	15
55	RNA profiling reveals familial aggregation of molecular subtypes in non-BRCA1/2 breast cancer families. <i>BMC Medical Genomics</i> , <b>2014</b> , 7, 9	3.7	15
54	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. <i>Breast Cancer Research and Treatment</i> , <b>2017</b> , 161, 117-134	4.4	15
53	Intragenic duplication: a novel mutational mechanism in hereditary pancreatitis. <i>Pancreas</i> , <b>2011</b> , 40, 540	<b>)-£</b> 6	14
52	Interferon stimulates the expression of 2',5'-oligoadenylate synthetase and MHC class I antigens in insulin-producing cells. <i>Journal of Interferon Research</i> , <b>1991</b> , 11, 255-60		14
51	Awareness of endometrial cancer risk and compliance with screening in hereditary nonpolyposis colorectal cancer. <i>Obstetrics and Gynecology</i> , <b>2012</b> , 120, 1005-12	4.9	14
50	The results of gynecologic surveillance in families with hereditary nonpolyposis colorectal cancer. <i>Gynecologic Oncology</i> , <b>2014</b> , 133, 526-30	4.9	13
49	Novel germline c-MET mutation in a family with hereditary papillary renal carcinoma. <i>Familial Cancer</i> , <b>2012</b> , 11, 535-7	3	13

## (2008-2018)

47	Oral Contraceptive Use and Breast Cancer Risk: Retrospective and Prospective Analyses From a BRCA1 and BRCA2 Mutation Carrier Cohort Study. <i>JNCI Cancer Spectrum</i> , <b>2018</b> , 2, pky023	4.6	13
46	Identification of eight novel SDHB, SDHC, SDHD germline variants in Danish pheochromocytoma/paraganglioma patients. <i>Hereditary Cancer in Clinical Practice</i> , <b>2016</b> , 14, 13	2.3	12
45	Genetic screening of the FLCN gene identify six novel variants and a Danish founder mutation. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 151-157	4.3	12
44	Germline RAD51B truncating mutation in a family with cutaneous melanoma. <i>Familial Cancer</i> , <b>2015</b> , 14, 337-40	3	11
43	Pallister-Killian syndrome: Multiband FISH of tetrasomy 12p. <i>Pediatric Dermatology</i> , <b>2006</b> , 23, 378-81	1.9	11
42	Pediatric cancer families' participation in whole-genome sequencing research in Denmark: Parent perspectives. <i>European Journal of Cancer Care</i> , <b>2018</b> , 27, e12877	2.4	10
41	Germline Variation at CDKN2A and Associations with Nevus Phenotypes among Members of Melanoma Families. <i>Journal of Investigative Dermatology</i> , <b>2017</b> , 137, 2606-2612	4.3	10
40	Incidence, etiology and prognosis of first-time acute pancreatitis in young patients: a population-based cohort study. <i>Pancreatology</i> , <b>2010</b> , 10, 453-61	3.8	10
39	The Influence of Number and Timing of Pregnancies on Breast Cancer Risk for Women With or Mutations. <i>JNCI Cancer Spectrum</i> , <b>2018</b> , 2, pky078	4.6	10
38	Identification of a breast cancer family double heterozygote for RAD51C and BRCA2 gene mutations. <i>Familial Cancer</i> , <b>2015</b> , 14, 129-33	3	9
37	Alcohol Consumption, Cigarette Smoking, and Risk of Breast Cancer for and Mutation Carriers: Results from The BRCA1 and BRCA2 Cohort Consortium. <i>Cancer Epidemiology Biomarkers and Prevention</i> , <b>2020</b> , 29, 368-378	4	9
36	Phenotypic and Histopathological Tumor Characteristics According to CDKN2A Mutation Status among Affected Members of Melanoma Families. <i>Journal of Investigative Dermatology</i> , <b>2016</b> , 136, 1066	-10369	9
35	Estimating CDKN2A mutation carrier probability among global familial melanoma cases using GenoMELPREDICT. <i>Journal of the American Academy of Dermatology</i> , <b>2019</b> , 81, 386-394	4.5	9
34	A new family with a homozygous nonsense variant in further delineated the clinical phenotype of -associated polyposis. <i>Human Genome Variation</i> , <b>2019</b> , 6, 46	1.8	9
33	A missense mutation in exon 13 in BRCA2, c.7235G>A, results in skipping of exon 13. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2006</b> , 10, 116-20		9
32	Oral contraceptive use and ovarian cancer risk for BRCA1/2 mutation carriers: an international cohort study. <i>American Journal of Obstetrics and Gynecology</i> , <b>2021</b> , 225, 51.e1-51.e17	6.4	9
31	Biomedical informatics as support to individual healthcare in hereditary colon cancer: the Danish HNPCC system. <i>Human Mutation</i> , <b>2011</b> , 32, 551-6	4.7	8
30	Familial pancreatic cancer. Scandinavian Journal of Gastroenterology, 2008, 43, 387-97	2.4	8

29	Functional characterization of rare missense mutations in MLH1 and MSH2 identified in Danish colorectal cancer patients. <i>Familial Cancer</i> , <b>2009</b> , 8, 489-500	3	7
28	Is the SPINK1 variant p.N34S overrepresented in patients with acute pancreatitis?. <i>European Journal of Gastroenterology and Hepatology</i> , <b>2012</b> , 24, 309-15	2.2	7
27	Medullary thyroid cancer: RET testing of an archival material. <i>European Archives of Oto-Rhino-Laryngology</i> , <b>2010</b> , 267, 613-7	3.5	7
26	Fine-Scale Mapping at 9p22.2 Identifies Candidate Causal Variants That Modify Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. <i>PLoS ONE</i> , <b>2016</b> , 11, e0158801	3.7	7
25	Fluorescence in situ hybridization of old G-banded and mounted chromosome preparations. <i>Cancer Genetics and Cytogenetics</i> , <b>1997</b> , 98, 9-15		6
24	Exploring the hereditary background of renal cancer in Denmark. <i>PLoS ONE</i> , <b>2019</b> , 14, e0215725	3.7	5
23	High accuracy of family history of melanoma in Danish melanoma cases. Familial Cancer, 2015, 14, 609-7	133	5
22	Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. <i>Lancet Oncology, The</i> , <b>2021</b> , 22, 1014-1022	21.7	5
21	Classification of the spliceogenic c.4096+3A>G variant as likely benign based on cosegregation data and identification of a healthy homozygous carrier. <i>Clinical Case Reports (discontinued)</i> , <b>2017</b> , 5, 876-879	9 <sup>0.7</sup>	4
20	Screening of 1331 Danish breast and/or ovarian cancer families identified 40 novel BRCA1 and BRCA2 mutations. <i>Familial Cancer</i> , <b>2011</b> , 10, 207-12	3	4
19	A BRCA2 mutation incorrectly mapped in the original BRCA2 reference sequence, is a common West Danish founder mutation disrupting mRNA splicing. <i>Breast Cancer Research and Treatment</i> , <b>2011</b> , 128, 179-85	4.4	4
18	Tumour-infiltrating CD4-, CD8- and FOXP3-positive immune cells as predictive markers of mortality in BRCA1- and BRCA2-associated breast cancer. <i>British Journal of Cancer</i> , <b>2021</b> , 125, 1388-1398	8.7	4
17	A putative Lynch syndrome family carrying MSH2 and MSH6 variants of uncertain significance-functional analysis reveals the pathogenic one. <i>Familial Cancer</i> , <b>2011</b> , 10, 515-20	3	3
16	New Pathogenic Germline Variants in Very Early Onset and Familial Colorectal Cancer Patients. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 566266	4.5	3
15	Cowden Syndrome and Concomitant Pulmonary Neuroendocrine Tumor: A Presentation of Two Cases. <i>Case Reports in Medicine</i> , <b>2015</b> , 2015, 265786	0.7	2
14	Risks of breast and ovarian cancer for women harboring pathogenic missense variants in BRCA1 and BRCA2 compared with those harboring protein truncating variants <i>Genetics in Medicine</i> , <b>2021</b> ,	8.1	2
13	The predictive ability of the 313 variant-based polygenic risk score for contralateral breast cancer risk prediction in women of European ancestry with a heterozygous BRCA1 or BRCA2 pathogenic variant. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1726-1737	8.1	2
12	Direct to consumer genetic testing in Denmark-public knowledge, use, and attitudes. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 851-860	5.3	2

## LIST OF PUBLICATIONS

11	KLHDC8B Is a Novel, Mitotically-Regulated Classical Hodgkin Lymphoma Candidate Susceptibility Gene <i>Blood</i> , <b>2006</b> , 108, 473-473	2.2	1	
10	Clinical characteristics and registry-validated extended pedigrees of germline TP53 mutation carriers in Denmark. <i>PLoS ONE</i> , <b>2018</b> , 13, e0190050	3.7	1	
9	Genomic profiling of tumors from patients with germline BRCA mutations <i>Journal of Clinical Oncology</i> , <b>2018</b> , 36, 1533-1533	2.2	1	
8	Computational and cellular studies reveal structural destabilization and degradation of MLH1 variants in Lynch syndrome		1	
7	Breast cancer survival in Nordic BRCA2 mutation carriers-unconventional association with oestrogen receptor status. <i>British Journal of Cancer</i> , <b>2020</b> , 123, 1608-1615	8.7	1	
6	Selection criteria for assembling a pediatric cancer predisposition syndrome gene panel. <i>Familial Cancer</i> , <b>2021</b> , 20, 279-287	3	1	
5	Cohort profile and heritability assessment of familial pancreatic cancer: a nation-wide study. <i>Scandinavian Journal of Gastroenterology</i> , <b>2021</b> , 56, 965-971	2.4	1	
4	A rare missense variant in interrupts splicing and causes AFAP in two Danish families. <i>Hereditary Cancer in Clinical Practice</i> , <b>2020</b> , 18, 8	2.3	1	
3	Whole genome sequencing identifies rare germline variants enriched in cancer related genes in first degree relatives of familial pancreatic cancer patients. <i>Clinical Genetics</i> , <b>2021</b> , 100, 551-562	4	1	
2	Prevalence of Pathogenic Germline Variants in Young Individuals Thyroidectomised Due to Goitre - A National Danish Cohort. <i>Frontiers in Endocrinology</i> , <b>2021</b> , 12, 727970	5.7	1	
1	Population-based whole-genome sequencing with constrained gene analysis identifies predisposing germline variants in children with central nervous system tumors		1	