

# Paivi T Peltomäki

## List of Publications by Year in descending order

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69  
papers

8,225  
citations

172457

29  
h-index

102487

66  
g-index

69  
all docs

69  
docs citations

69  
times ranked

6573  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutation of a <i>mutL</i> Homolog in Hereditary Colon Cancer. <i>Science</i> , 1994, 263, 1625-1629.	12.6	1,821
2	Analysis of mismatch repair genes in hereditary non-polyposis colorectal cancer patients. <i>Nature Medicine</i> , 1996, 2, 169-174.	30.7	892
3	Mismatch repair gene defects in sporadic colorectal cancers with microsatellite instability. <i>Nature Genetics</i> , 1995, 9, 48-55.	21.4	759
4	Role of DNA Mismatch Repair Defects in the Pathogenesis of Human Cancer. <i>Journal of Clinical Oncology</i> , 2003, 21, 1174-1179.	1.6	626
5	Mutations Associated with HNPCC Predisposition – Update of ICG-HNPCC/INSiGHT Mutation Database. <i>Disease Markers</i> , 2004, 20, 269-276.	1.3	416
6	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. <i>Nature Genetics</i> , 2014, 46, 107-115.	21.4	410
7	Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. <i>Nature Genetics</i> , 1994, 8, 405-410.	21.4	304
8	Founding mutations and Alu-mediated recombination in hereditary colon cancer. <i>Nature Medicine</i> , 1995, 1, 1203-1206.	30.7	275
9	Genetic and Epigenetic Modification of MLH1 Accounts for a Major Share of Microsatellite-Unstable Colorectal Cancers. <i>American Journal of Pathology</i> , 2000, 156, 1773-1779.	3.8	255
10	Lynch Syndrome Genes. <i>Familial Cancer</i> , 2005, 4, 227-232.	1.9	219
11	Germline Mutation of RPS20, Encoding a Ribosomal Protein, Causes Predisposition to Hereditary Nonpolyposis Colorectal Carcinoma Without DNA Mismatch Repair Deficiency. <i>Gastroenterology</i> , 2014, 147, 595-598.e5.	1.3	143
12	Update on Lynch syndrome genomics. <i>Familial Cancer</i> , 2016, 15, 385-393.	1.9	127
13	Molecular Analysis of Familial Endometrial Carcinoma: A Manifestation of Hereditary Nonpolyposis Colorectal Cancer or a Separate Syndrome?. <i>Journal of Clinical Oncology</i> , 2005, 23, 4609-4616.	1.6	125
14	Predictive genetic testing for hereditary non-polyposis colorectal cancer: Uptake and long-term satisfaction. <i>International Journal of Cancer</i> , 2000, 89, 44-50.	5.1	116
15	Sporadic gastric carcinomas with microsatellite instability display a particular clinicopathologic profile. <i>International Journal of Cancer</i> , 1995, 64, 32-36.	5.1	110
16	Molecular mapping of the putative gonadoblastoma locus on the Y chromosome. <i>Genes Chromosomes and Cancer</i> , 1995, 14, 210-214.	2.8	101
17	MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. , 1997, 18, 269-278.		99
18	Deletion of 1p loci and microsatellite instability in colorectal polyps. <i>Genes Chromosomes and Cancer</i> , 1995, 14, 182-188.	2.8	90

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19	Altered Expression of MLH1, MSH2, and MSH6 in Predisposition to Hereditary Nonpolyposis Colorectal Cancer. <i>Journal of Clinical Oncology</i> , 2003, 21, 3629-3637.	1.6	88
20	Large genomic rearrangements and germline epimutations in Lynch syndrome. <i>International Journal of Cancer</i> , 2009, 124, 2333-2340.	5.1	80
21	Comprehensive characterization of HNPCC-related colorectal cancers reveals striking molecular features in families with no germline mismatch repair gene mutations. <i>Oncogene</i> , 2005, 24, 1542-1551.	5.9	79
22	Epigenetic Signatures of Familial Cancer Are Characteristic of Tumor Type and Family Category. <i>Cancer Research</i> , 2008, 68, 4597-4605.	0.9	79
23	Mismatch repair genes and mononucleotide tracts as mutation targets in colorectal tumors with different degrees of microsatellite instability. <i>Oncogene</i> , 1998, 17, 157-163.	5.9	68
24	Mutations and epimutations in the origin of cancer. <i>Experimental Cell Research</i> , 2012, 318, 299-310.	2.6	68
25	Microsatellite instability in cervical and endometrial carcinomas. <i>International Journal of Cancer</i> , 1997, 70, 499-501.	5.1	64
26	Associations of Pathogenic Variants in MLH1, MSH2, and MSH6 With Risk of Colorectal Adenomas and Tumors and With Somatic Mutations in Patients With Lynch Syndrome. <i>Gastroenterology</i> , 2020, 158, 1326-1333.	1.3	60
27	Biallelic germline nonsense variant of MLH3 underlies polyposis predisposition. <i>Genetics in Medicine</i> , 2019, 21, 1868-1873.	2.4	39
28	Mutation sharing, predominant involvement of the MLH1 gene and description of four novel mutations in hereditary nonpolyposis colorectal cancer. <i>Human Mutation</i> , 1998, 11, 482-483.	2.5	33
29	Epigenetic analysis of sporadic and Lynch-associated ovarian cancers reveals histology-specific patterns of DNA methylation. <i>Epigenetics</i> , 2014, 9, 1577-1587.	2.7	32
30	Somatic <i>FGF9</i> mutations in colorectal and endometrial carcinomas associated with membranous $\beta$ -catenin. <i>Human Mutation</i> , 2008, 29, 390-397.	2.5	31
31	Distinct molecular profiles in Lynch syndrome-associated and sporadic ovarian carcinomas. <i>International Journal of Cancer</i> , 2013, 133, n/a-n/a.	5.1	29
32	Desmoid tumor patients carry an elevated risk of familial adenomatous polyposis. <i>Journal of Surgical Oncology</i> , 2016, 113, 209-212.	1.7	29
33	Converging endometrial and ovarian tumorigenesis in Lynch syndrome: Shared origin of synchronous carcinomas. <i>Gynecologic Oncology</i> , 2018, 150, 92-98.	1.4	29
34	Epidemiological, clinical and molecular characterization of Lynch-like syndrome: A population-based study. <i>International Journal of Cancer</i> , 2019, 145, 87-98.	5.1	28
35	Genetic Basis of Hereditary Nonpolyposis Colorectal Carcinoma (HNPCC). <i>Annals of Medicine</i> , 1994, 26, 215-219.	3.8	27
36	Molecular changes preceding endometrial and ovarian cancer: a study of consecutive endometrial specimens from Lynch syndrome surveillance. <i>Modern Pathology</i> , 2018, 31, 1291-1301.	5.5	25

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37	Oncogenic Potential of Bisphenol A and Common Environmental Contaminants in Human Mammary Epithelial Cells. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3735.	4.1	25
38	Distinct Genetic and Epigenetic Signatures of Colorectal Cancers According to Ethnic Origin. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2012, 21, 202-211.	2.5	24
39	DNA hypermethylation appears early and shows increased frequency with dysplasia in Lynch syndrome-associated colorectal adenomas and carcinomas. <i>Clinical Epigenetics</i> , 2015, 7, 71.	4.1	24
40	MicroRNA Methylation in Colorectal Cancer. <i>Advances in Experimental Medicine and Biology</i> , 2016, 937, 109-122.	1.6	24
41	Microsatellite instability and hereditary non-polyposis colon cancer. <i>Journal of Pathology</i> , 1995, 176, 329-330.	4.5	22
42	DNA methylation changes and somatic mutations as tumorigenic events in Lynch syndrome-associated adenomas retaining mismatch repair protein expression. <i>EBioMedicine</i> , 2019, 39, 280-291.	6.1	21
43	Identification of subgroup-specific miRNA patterns by epigenetic profiling of sporadic and Lynch syndrome-associated colorectal and endometrial carcinoma. <i>Clinical Epigenetics</i> , 2015, 7, 20.	4.1	20
44	Molecular cytogenetic study of patients with Pallister-Killian syndrome. <i>Human Genetics</i> , 1993, 91, 121-127.	3.8	19
45	Altered dosage of the sex chromosomes in human testicular cancer: A molecular genetic study. <i>International Journal of Cancer</i> , 1991, 47, 518-522.	5.1	18
46	Sequencing of Lynch syndrome tumors reveals the importance of epigenetic alterations. <i>Oncotarget</i> , 2017, 8, 108020-108030.	1.8	18
47	Mlh1 deficiency in normal mouse colon mucosa associates with chromosomally unstable colon cancer. <i>Carcinogenesis</i> , 2018, 39, 788-797.	2.8	18
48	Updates in the field of hereditary nonpolyposis colorectal cancer. <i>Expert Review of Gastroenterology and Hepatology</i> , 2020, 14, 707-720.	3.0	18
49	Thyroid Carcinomas That Occur in Familial Adenomatous Polyposis Patients Recurrently Harbor Somatic Variants in <i>APC</i> , <i>BRAF</i> , and <i>KMT2D</i> . <i>Thyroid</i> , 2020, 30, 380-388.	4.5	18
50	The Role of Chromosomal Instability and Epigenetics in Colorectal Cancers Lacking $\beta$ -Catenin/TCF Regulated Transcription. <i>Gastroenterology Research and Practice</i> , 2016, 2016, 1-11.	1.5	17
51	Pseudoexons provide a mechanism for allele-specific expression of <i>APC</i> in familial adenomatous polyposis. <i>Oncotarget</i> , 2016, 7, 70685-70698.	1.8	17
52	From <i>APC</i> to the genetics of hereditary and familial colon cancer syndromes. <i>Human Molecular Genetics</i> , 2021, 30, R206-R224.	2.9	15
53	Molecular studies of the sex chromosomes in human testicular cancer: Pronounced changes in X and Y chromosome dosage in some tumors. <i>Genes Chromosomes and Cancer</i> , 1989, 1, 42-47.	2.8	14
54	Restoring mismatch repair does not stop the formation of reciprocal translocations in the colon cancer cell line HCA7 but further destabilizes chromosome number. <i>Oncogene</i> , 2005, 24, 706-713.	5.9	14

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55	Methyltransferase expression and tumor suppressor gene methylation in sporadic and familial colorectal cancer. <i>Genes Chromosomes and Cancer</i> , 2015, 54, 776-787.	2.8	14
56	No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study. <i>Journal of Clinical Medicine</i> , 2021, 10, 2856.	2.4	11
57	Does breast carcinoma belong to the Lynch syndrome tumor spectrum? â€œ Somatic mutational profiles vs. ovarian and colorectal carcinomas. <i>Oncotarget</i> , 2020, 11, 1244-1256.	1.8	11
58	Somatic mutation profiles as molecular classifiers of ulcerative colitis-associated colorectal cancer. <i>International Journal of Cancer</i> , 2021, 148, 2997-3007.	5.1	10
59	Differential roles of EPS8 in carcinogenesis: Loss of protein expression in a subset of colorectal carcinoma and adenoma. <i>World Journal of Gastroenterology</i> , 2012, 18, 3896.	3.3	10
60	Gene fusions and oncogenic mutations in MLH1 deficient and BRAFV600E wild-type colorectal cancers. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2022, 480, 807-817.	2.8	9
61	Molecular Basis of Mismatch Repair Protein Deficiency in Tumors from Lynch Suspected Cases with Negative Germline Test Results. <i>Cancers</i> , 2020, 12, 1853.	3.7	8
62	Characterization of neoplastic and reactive cells in T-cell lymphomas with cytogenetic, surface marker, and DNA methods. <i>British Journal of Haematology</i> , 1989, 73, 68-75.	2.5	7
63	Chromosomal abnormality limited to T4 lymphocytes in a patient with Tâ€cell chronic lymphocytic leukaemia. <i>European Journal of Haematology</i> , 1990, 45, 52-59.	2.2	7
64	Body Weight, Physical Activity, and Risk of Cancer in Lynch Syndrome. <i>Cancers</i> , 2021, 13, 1849.	3.7	6
65	MSH2 and MLH1 mutations in sporadic replication errorâ€positive colorectal carcinoma as assessed by twoâ€dimensional DNA electrophoresis. <i>Genes Chromosomes and Cancer</i> , 1997, 18, 269-278.	2.8	4
66	Immunoprofiles and DNA Methylation of Inflammatory Marker Genes in Ulcerative Colitis-Associated Colorectal Tumorigenesis. <i>Biomolecules</i> , 2021, 11, 1440.	4.0	3
67	Testing for Lynch Syndrome in Endometrial Carcinoma: From Universal to Age-Selective MLH1 Methylation Analysis. <i>Cancers</i> , 2022, 14, 1348.	3.7	3
68	Tracking mutations in a family: Recognizing indicators of germline mutation in Lynch syndrome. <i>Current Colorectal Cancer Reports</i> , 2007, 3, 199-205.	0.5	0
69	Reply. <i>Gastroenterology</i> , 2015, 148, 259.	1.3	0