Lauri A Aaltonen

List of Publications by Citations

308 papers	35,805 citations	90 h-index	185 g-index
349 ext. papers	39,180 ext. citations	11 avg, IF	6.22 L-index

#	Paper	IF	Citations
308	Clues to the pathogenesis of familial colorectal cancer. *Science*, **1993**, 260, 812-6	33.3	2342
307	Mutations of a mutS homolog in hereditary nonpolyposis colorectal cancer. *Cell*, **1993**, 75, 1215-25	56.2	1954
306	A serine/threonine kinase gene defective in Peutz-Jeghers syndrome. *Nature*, **1998**, 391, 184-7	50.4	1284
305	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. *Nature Genetics*, **2002**, 30, 406-10	36.3	1191
304	Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology*, **2000**, 118, 829-34	13.3	1098
303	Incidence of hereditary nonpolyposis colorectal cancer and the feasibility of molecular screening for the disease. *New England Journal of Medicine*, **1998**, 338, 1481-7	59.2	928
302	Cancer risk in mutation carriers of DNA-mismatch-repair genes. *International Journal of Cancer*, **1999**, 81, 214-8	7.5	905
301	Genetic mapping of a locus predisposing to human colorectal cancer. *Science*, **1993**, 260, 810-2	33.3	762
300	Mutations in the SMAD4/DPC4 gene in juvenile polyposis. *Science*, **1998**, 280, 1086-8	33.3	759
299	Multiple colorectal adenomas, classic adenomatous polyposis, and germ-line mutations in MYH. *New England Journal of Medicine*, **2003**, 348, 791-9	59.2	721
298	Inherited susceptibility to uterine leiomyomas and renal cell cancer. *Proceedings of the National Academy of Sciences of the United States of America*, **2001**, 98, 3387-92	11.5	515
297	Life-time risk of different cancers in hereditary non-polyposis colorectal cancer (HNPCC) syndrome. *International Journal of Cancer*, **1995**, 64, 430-3	7.5	503
296	A genome-wide association study identifies colorectal cancer susceptibility loci on chromosomes 10p14 and 8q23.3. *Nature Genetics*, **2008**, 40, 623-30	36.3	463
295	Pituitary adenoma predisposition caused by germline mutations in the AIP gene. *Science*, **2006**, 312, 1228-30	33.3	463
294	Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer. *Nature Genetics*, **2008**, 40, 1426-35	36.3	457
293	MED12, the mediator complex subunit 12 gene, is mutated at high frequency in uterine leiomyomas. *Science*, **2011**, 334, 252-5	33.3	449
292	Loss-of-function mutations in PPAR gamma associated with human colon cancer. *Molecular Cell*, **1999**, 3, 799-804	17.6	438

291	The common colorectal cancer predisposition SNP rs6983267 at chromosome 8q24 confers potential to enhanced Wnt signaling. *Nature Genetics*, **2009**, 41, 885-90	36.3	422
290	Population-based molecular detection of hereditary nonpolyposis colorectal cancer. *Journal of Clinical Oncology*, **2000**, 18, 2193-200	2.2	414
289	Localization of a susceptibility locus for Peutz-Jeghers syndrome to 19p using comparative genomic hybridization and targeted linkage analysis. *Nature Genetics*, **1997**, 15, 87-90	36.3	385
288	A recurrent mutation in PALB2 in Finnish cancer families. *Nature*, **2007**, 446, 316-9	50.4	349
287	DNA methylation patterns in hereditary human cancers mimic sporadic tumorigenesis. *Human Molecular Genetics*, **2001**, 10, 3001-7	5.6	328
286	Identification of Lynch syndrome among patients with colorectal cancer. *JAMA - Journal of the American Medical Association*, **2012**, 308, 1555-65	27.4	323
285	Metastasis-Associated Gene Expression Changes Predict Poor Outcomes in Patients with Dukes Stage B and C Colorectal Cancer. *Clinical Cancer Research*, **2009**, 15, 7642-7651	12.9	320
284	A TARBP2 mutation in human cancer impairs microRNA processing and DICER1 function. *Nature Genetics*, **2009**, 41, 365-70	36.3	317
283	A role for mitochondrial enzymes in inherited neoplasia and beyond. *Nature Reviews Cancer*, **2003**, 3, 193-202	31.3	316
282	Cancer risk in hereditary nonpolyposis colorectal cancer syndrome: later age of onset. *Gastroenterology*, **2005**, 129, 415-21	13.3	312
281	Early-onset renal cell carcinoma as a novel extraparaganglial component of SDHB-associated heritable paraganglioma. *American Journal of Human Genetics*, **2004**, 74, 153-9	11	311
280	Better survival rates in patients with MLH1-associated hereditary colorectal cancer. *Gastroenterology*, **1996**, 110, 682-7	13.3	302
279	Meta-analysis of three genome-wide association studies identifies susceptibility loci for colorectal cancer at 1q41, 3q26.2, 12q13.13 and 20q13.33. *Nature Genetics*, **2010**, 42, 973-7	36.3	301
278	Microsatellite instability is a favorable prognostic indicator in patients with colorectal cancer receiving chemotherapy. *Gastroenterology*, **2000**, 119, 921-8	13.3	291
277	CTCF/cohesin-binding sites are frequently mutated in cancer. *Nature Genetics*, **2015**, 47, 818-21	36.3	286
276	Mutations in BRIP1 confer high risk of ovarian cancer. *Nature Genetics*, **2011**, 43, 1104-7	36.3	285
275	Molecular staging for survival prediction of colorectal cancer patients. *Journal of Clinical Oncology*, **2005**, 23, 3526-35	2.2	285
274	Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. *Nature Genetics*, **1994**, 8, 405-10	36.3	281

273	Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer Syndrome: Later Age of Onset. *Gastroenterology*, **2005**, 129, 415-421	13.3	263
272	Clinical characteristics and therapeutic responses in patients with germ-line AIP mutations and pituitary adenomas: an international collaborative study. *Journal of Clinical Endocrinology and Metabolism*, **2010**, 95, E373-83	5.6	259
271	BRAF screening as a low-cost effective strategy for simplifying HNPCC genetic testing. *Journal of Medical Genetics*, **2004**, 41, 664-8	5.8	256
270	Founding mutations and Alu-mediated recombination in hereditary colon cancer. *Nature Medicine*, **1995**, 1, 1203-6	50.5	254
269	Familial isolated pituitary adenomas (FIPA) and the pituitary adenoma predisposition due to mutations in the aryl hydrocarbon receptor interacting protein (AIP) gene. *Endocrine Reviews*, **2013**, 34, 239-77	27.2	232
268	Germline CDKN1B/p27Kip1 mutation in multiple endocrine neoplasia. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 3321-5	5.6	230
267	A truncating mutation of HDAC2 in human cancers confers resistance to histone deacetylase inhibition. *Nature Genetics*, **2006**, 38, 566-9	36.3	228
266	Characterization of uterine leiomyomas by whole-genome sequencing. *New England Journal of Medicine*, **2013**, 369, 43-53	59.2	223
265	Ten years after mutation testing for Lynch syndrome: cancer incidence and outcome in mutation-positive and mutation-negative family members. *Journal of Clinical Oncology*, **2009**, 27, 4793-7	2.2	209
264	Germline mutations in BMPR1A/ALK3 cause a subset of cases of juvenile polyposis syndrome and of Cowden and Bannayan-Riley-Ruvalcaba syndromes. *American Journal of Human Genetics*, **2001**, 69, 704-11	11	208
263	The prevalence of MADH4 and BMPR1A mutations in juvenile polyposis and absence of BMPR2, BMPR1B, and ACVR1 mutations. *Journal of Medical Genetics*, **2004**, 41, 484-91	5.8	190
262	Molecular classification of patients with unexplained hamartomatous and hyperplastic polyposis. *JAMA - Journal of the American Medical Association*, **2005**, 294, 2465-73	27.4	189
261	Common variation near CDKN1A, POLD3 and SHROOM2 influences colorectal cancer risk. *Nature Genetics*, **2012**, 44, 770-6	36.3	184
260	Aberrant succination of proteins in fumarate hydratase-deficient mice and HLRCC patients is a robust biomarker of mutation status. *Journal of Pathology*, **2011**, 225, 4-11	9.4	184
259	The DNA repair gene MBD4 (MED1) is mutated in human carcinomas with microsatellite instability. *Nature Genetics*, **1999**, 23, 266-8	36.3	183
258	Differential gene expression in colon cancer of the caecum versus the sigmoid and rectosigmoid. *Gut*, **2005**, 54, 374-84	19.2	179
257	Mice lacking a Myc enhancer that includes human SNP rs6983267 are resistant to intestinal tumors. *Science*, **2012**, 338, 1360-3	33.3	171
256	Multiple common susceptibility variants near BMP pathway loci GREM1, BMP4, and BMP2 explain part of the missing heritability of colorectal cancer. *PLoS Genetics*, **2011**, 7, e1002105	6	169

255	Familial cutaneous leiomyomatosis is a two-hit condition associated with renal cell cancer of characteristic histopathology. *American Journal of Pathology*, **2001**, 159, 825-9	5.8	162
254	PTEN mutational spectra, expression levels, and subcellular localization in microsatellite stable and unstable colorectal cancers. *American Journal of Pathology*, **2002**, 161, 439-47	5.8	161
253	Gene expression in colorectal cancer. *Cancer Research*, **2002**, 62, 4352-63	10.1	161
252	Increased risk of cancer in patients with fumarate hydratase germline mutation. *Journal of Medical Genetics*, **2006**, 43, 523-6	5.8	155
251	SMAD4 as a prognostic marker in colorectal cancer. *Clinical Cancer Research*, **2005**, 11, 2606-11	12.9	149
250	Epigenetic inactivation of LKB1 in primary tumors associated with the Peutz-Jeghers syndrome. *Oncogene*, **2000**, 19, 164-8	9.2	149
249	Molecular diagnosis of pituitary adenoma predisposition caused by aryl hydrocarbon receptor-interacting protein gene mutations. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 4101-5	11.5	147
248	Explaining the familial colorectal cancer risk associated with mismatch repair (MMR)-deficient and MMR-stable tumors. *Clinical Cancer Research*, **2007**, 13, 356-61	12.9	145
247	Biallelic inactivation of fumarate hydratase (FH) occurs in nonsyndromic uterine leiomyomas but is rare in other tumors. *American Journal of Pathology*, **2004**, 164, 17-22	5.8	143
246	Screening E-cadherin in gastric cancer families reveals germline mutations only in hereditary diffuse gastric cancer kindred. *Human Mutation*, **2002**, 19, 510-7	4.7	142
245	Transcription factor PROX1 induces colon cancer progression by promoting the transition from benign to highly dysplastic phenotype. *Cancer Cell*, **2008**, 13, 407-19	24.3	140
244	BRAF-V600E is not involved in the colorectal tumorigenesis of HNPCC in patients with functional MLH1 and MSH2 genes. *Oncogene*, **2005**, 24, 3995-8	9.2	128
243	Features of gastric cancer in hereditary non-polyposis colorectal cancer syndrome. *International Journal of Cancer*, **1997**, 74, 551-5	7.5	124
242	Low-level microsatellite instability in most colorectal carcinomas. *Cancer Research*, **2002**, 62, 1166-70	10.1	122
241	Serrated carcinomas form a subclass of colorectal cancer with distinct molecular basis. *Oncogene*, **2007**, 26, 312-20	9.2	118
240	Proportion and phenotype of MYH-associated colorectal neoplasia in a population-based series of Finnish colorectal cancer patients. *American Journal of Pathology*, **2003**, 163, 827-32	5.8	117
239	Integrated data analysis reveals uterine leiomyoma subtypes with distinct driver pathways and biomarkers. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, 1315-20	11.5	116
238	Mutations and impaired function of LKB1 in familial and non-familial Peutz-Jeghers syndrome and a sporadic testicular cancer. *Human Molecular Genetics*, **1999**, 8, 45-51	5.6	114

237	Genomics of uterine leiomyomas: insights from high-throughput sequencing. *Fertility and Sterility*, **2014**, 102, 621-9	4.8	112
236	LKB1 exonic and whole gene deletions are a common cause of Peutz-Jeghers syndrome. *Journal of Medical Genetics*, **2006**, 43, e18	5.8	112
235	Genetic reconstruction of individual colorectal tumor histories. *Proceedings of the National Academy of Sciences of the United States of America*, **2000**, 97, 1236-41	11.5	111
234	Few FH mutations in sporadic counterparts of tumor types observed in hereditary leiomyomatosis and renal cell cancer families. *Cancer Research*, **2002**, 62, 4554-7	10.1	111
233	Development of colorectal tumors in colonoscopic surveillance in Lynch syndrome. *Gastroenterology*, **2007**, 133, 1093-8	13.3	110
232	LKB1 somatic mutations in sporadic tumors. *American Journal of Pathology*, **1999**, 154, 677-81	5.8	110
231	Gene-expression profiling predicts recurrence in Dukes' C colorectal cancer. *Gastroenterology*, **2005**, 129, 874-84	13.3	106
230	Loss of SUFU function in familial multiple meningioma. *American Journal of Human Genetics*, **2012**, 91, 520-6	11	103
229	Induction of cyclooxygenase-2 in a mouse model of Peutz-Jeghers polyposis. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 12327-32	11.5	103
228	Distinct patterns of KRAS mutations in colorectal carcinomas according to germline mismatch repair defects and hMLH1 methylation status. *Human Molecular Genetics*, **2004**, 13, 2303-11	5.6	102
227	Frequent loss of SMAD4/DPC4 protein in colorectal cancers. *Gut*, **2002**, 51, 56-9	19.2	102
226	Uterine leiomyoma-linked MED12 mutations disrupt mediator-associated CDK activity. *Cell Reports*, **2014**, 7, 654-60	10.6	99
225	The MDM2 promoter polymorphism SNP309T-->G and the risk of uterine leiomyosarcoma, colorectal cancer, and squamous cell carcinoma of the head and neck. *Journal of Medical Genetics*, **2005**, 42, 694-8	5.8	97
224	KSHV-initiated notch activation leads to membrane-type-1 matrix metalloproteinase-dependent lymphatic endothelial-to-mesenchymal transition. *Cell Host and Microbe*, **2011**, 10, 577-90	23.4	96
223	Cumulative impact of common genetic variants and other risk factors on colorectal cancer risk in 42,103 individuals. *Gut*, **2013**, 62, 871-81	19.2	95
222	Epigenetic phenotypes distinguish microsatellite-stable and -unstable colorectal cancers. *Proceedings of the National Academy of Sciences of the United States of America*, **1999**, 96, 12661-6	11.5	95
221	The MDM2 promoter SNP285C/309G haplotype diminishes Sp1 transcription factor binding and reduces risk for breast and ovarian cancer in Caucasians. *Cancer Cell*, **2011**, 19, 273-82	24.3	94
220	Somatic MED12 mutations in uterine leiomyosarcoma and colorectal cancer. *British Journal of Cancer*, **2012**, 107, 1761-5	8.7	91

219	A large-scale meta-analysis to refine colorectal cancer risk estimates associated with MUTYH variants. *British Journal of Cancer*, **2010**, 103, 1875-84	8.7	91
218	The role of hypermethylation of the hMLH1 promoter region in HNPCC versus MSI+ sporadic colorectal cancers. *Journal of Medical Genetics*, **2000**, 37, 588-92	5.8	89
217	Dysregulation of the transcription factors SOX4, CBFB and SMARCC1 correlates with outcome of colorectal cancer. *British Journal of Cancer*, **2009**, 100, 511-23	8.7	88
216	Microsatellite instability in adenomas as a marker for hereditary nonpolyposis colorectal cancer. *American Journal of Pathology*, **1999**, 155, 1849-53	5.8	84
215	Does MSI-low exist?. *Journal of Pathology*, **2002**, 197, 6-13	9.4	82
214	Association analyses identify 31 new risk loci for colorectal cancer susceptibility. *Nature Communications*, **2019**, 10, 2154	17.4	81
213	MDM2 SNP309 accelerates colorectal tumour formation in women. *Journal of Medical Genetics*, **2006**, 43, 950-2	5.8	81
212	SMAD4 levels and response to 5-fluorouracil in colorectal cancer. *Clinical Cancer Research*, **2005**, 11, 6311-6	10.9	79
211	Candidate driver genes in microsatellite-unstable colorectal cancer. *International Journal of Cancer*, **2012**, 130, 1558-66	7.5	78
210	MED12 exon 2 mutations are common in uterine leiomyomas from South African patients. *Oncotarget*, **2011**, 2, 966-9	3.3	78
209	Somatic microsatellite mutations as molecular tumor clocks. *Nature Medicine*, **1996**, 2, 676-81	50.5	77
208	Close linkage to chromosome 3p and conservation of ancestral founding haplotype in hereditary nonpolyposis colorectal cancer families. *Proceedings of the National Academy of Sciences of the United States of America*, **1994**, 91, 6054-8	11.5	77
207	Colibactin DNA-damage signature indicates mutational impact in colorectal cancer. *Nature Medicine* , **2020**, 26, 1063-1069	50.5	76
206	MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis **1997**, 18, 269-278		76
205	p53 codon 72 and MDM2 SNP309 polymorphisms and age of colorectal cancer onset in Lynch syndrome. *Clinical Cancer Research*, **2005**, 11, 6840-4	12.9	74
204	No support for endoscopic surveillance for gastric cancer in hereditary non-polyposis colorectal cancer. *Scandinavian Journal of Gastroenterology*, **2002**, 37, 574-7	2.4	74
203	Mechanisms of inactivation of the receptor tyrosine kinase EPHB2 in colorectal tumors. *Cancer Research*, **2005**, 65, 10170-3	10.1	73
202	Segregation of a missense variant in enteric smooth muscle actin Β2 with autosomal dominant familial visceral myopathy. *Gastroenterology*, **2012**, 143, 1482-1491.e3	13.3	71

201	Analysis of fumarate hydratase mutations in a population-based series of early onset uterine leiomyosarcoma patients. *International Journal of Cancer*, **2006**, 119, 283-7	7.5	69
200	EPHB4 and survival of colorectal cancer patients. *Cancer Research*, **2006**, 66, 8943-8	10.1	69
199	Allelic variation at the 8q23.3 colorectal cancer risk locus functions as a cis-acting regulator of EIF3H. *PLoS Genetics*, **2010**, 6, e1001126	6	68
198	HOXB13 G84E mutation in Finland: population-based analysis of prostate, breast, and colorectal cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2013**, 22, 452-60	4	67
197	DNA copy-number alterations underlie gene expression differences between microsatellite stable and unstable colorectal cancers. *Clinical Cancer Research*, **2008**, 14, 8061-9	12.9	67
196	Aryl hydrocarbon receptor interacting protein (AIP) gene mutation analysis in children and adolescents with sporadic pituitary adenomas. *Clinical Endocrinology*, **2008**, 69, 621-7	3.4	66
195	Large genomic deletions in AIP in pituitary adenoma predisposition. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 4146-51	5.6	66
194	SMAD genes in juvenile polyposis. *Genes Chromosomes and Cancer*, **1999**, 26, 54-61	5	66
193	Exome sequencing reveals germline NPAT mutation as a candidate risk factor for Hodgkin lymphoma. *Blood*, **2011**, 118, 493-8	2.2	65
192	Mismatch repair genes and mononucleotide tracts as mutation targets in colorectal tumors with different degrees of microsatellite instability. *Oncogene*, **1998**, 17, 157-63	9.2	65
191	Unregulated smooth-muscle myosin in human intestinal neoplasia. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 5513-8	11.5	64
190	Allelic imbalance at rs6983267 suggests selection of the risk allele in somatic colorectal tumor evolution. *Cancer Research*, **2008**, 68, 14-7	10.1	64
189	Whole-Genome Sequencing of Growth Hormone (GH)-Secreting Pituitary Adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 3918-27	5.6	62
188	Gene expression signatures for colorectal cancer microsatellite status and HNPCC. *British Journal of Cancer*, **2005**, 92, 2240-8	8.7	62
187	Mutations in the circadian gene CLOCK in colorectal cancer. *Molecular Cancer Research*, **2010**, 8, 952-60	6.6	60
186	Mice with inactivation of aryl hydrocarbon receptor-interacting protein (Aip) display complete penetrance of pituitary adenomas with aberrant ARNT expression. *American Journal of Pathology*, **2010**, 177, 1969-76	5.8	58
185	Comprehensive analysis of SMAD4 mutations and protein expression in juvenile polyposis: evidence for a distinct genetic pathway and polyp morphology in SMAD4 mutation carriers. *American Journal of Pathology*, **2001**, 159, 1293-300	5.8	58
184	Eleven candidate susceptibility genes for common familial colorectal cancer. *PLoS Genetics*, **2013**, 9, e1003876		57

183	Refinement of the basis and impact of common 11q23.1 variation to the risk of developing colorectal cancer. *Human Molecular Genetics*, **2008**, 17, 3720-7	5.6	57
182	Comparison of serous and mucinous ovarian carcinomas: distinct pattern of allelic loss at distal 8p and expression of transcription factor GATA-4. *Laboratory Investigation*, **2001**, 81, 517-26	5.9	57
181	Pro-inflammatory fatty acid profile and colorectal cancer risk: A Mendelian randomisation analysis. *European Journal of Cancer*, **2017**, 84, 228-238	7.5	56
180	MED12 exon 2 mutations in histopathological uterine leiomyoma variants. *European Journal of Human Genetics*, **2013**, 21, 1300-3	5.3	55
179	CHEK2 I157T associates with familial and sporadic colorectal cancer. *Journal of Medical Genetics*, **2006**, 43, e34	5.8	55
178	The I1307K polymorphism of the APC gene in colorectal cancer. *Gastroenterology*, **1999**, 116, 58-63	13.3	55
177	Expression profiling in progressive stages of fumarate-hydratase deficiency: the contribution of metabolic changes to tumorigenesis. *Cancer Research*, **2010**, 70, 9153-65	10.1	54
176	Distinct expression profile in fumarate-hydratase-deficient uterine fibroids. *Human Molecular Genetics*, **2006**, 15, 97-103	5.6	54
175	Subsequent primary malignancies after endometrial carcinoma and ovarian carcinoma. *Cancer*, **2003**, 97, 2432-9	6.4	53
174	Genomic profile of pseudomyxoma peritonei analyzed using next-generation sequencing and immunohistochemistry. *International Journal of Cancer*, **2015**, 136, E282-9	7.5	52
173	The expression of AIP-related molecules in elucidation of cellular pathways in pituitary adenomas. *American Journal of Pathology*, **2009**, 175, 2501-7	5.8	52
172	Differential expression of DHHC9 in microsatellite stable and instable human colorectal cancer subgroups. *British Journal of Cancer*, **2007**, 96, 1896-903	8.7	52
171	Colorectal adenoma and cancer divergence. Evidence of multilineage progression. *American Journal of Pathology*, **1999**, 154, 1815-24	5.8	52
170	MED12 mutation frequency in unselected sporadic uterine leiomyomas. *Fertility and Sterility*, **2014**, 102, 1137-42	4.8	51
169	Direct contact in inviting high-risk members of hereditary colon cancer families to genetic counselling and DNA testing. *Journal of Medical Genetics*, **2007**, 44, 732-8	5.8	51
168	Clusterin expression in normal mucosa and colorectal cancer. *Molecular and Cellular Proteomics*, **2007**, 6, 1039-48	7.6	51
167	Mendelian randomisation implicates hyperlipidaemia as a risk factor for colorectal cancer. *International Journal of Cancer*, **2017**, 140, 2701-2708	7.5	50
166	Lessons from functional analysis of genome-wide association studies. *Cancer Research*, **2013**, 73, 4180-4	10.1	50

165	Brush border myosin Ia has tumor suppressor activity in the intestine. *Proceedings of the National Academy of Sciences of the United States of America*, **2012**, 109, 1530-5	11.5	50
164	Diagnostic cancer genome sequencing and the contribution of germline variants. *Science*, **2013**, 339, 1559-62	33.3	49
163	Mutation analysis of aryl hydrocarbon receptor interacting protein (AIP) gene in colorectal, breast, and prostate cancers. *British Journal of Cancer*, **2007**, 96, 352-6	8.7	47
162	Prevalence of germline PTEN, BMPR1A, SMAD4, STK11, and ENG mutations in patients with moderate-load colorectal polyps. *Gastroenterology*, **2013**, 144, 1402-9, 1409.e1-5	13.3	46
161	Recessively inherited right atrial isomerism caused by mutations in growth/differentiation factor 1 (GDF1). *Human Molecular Genetics*, **2010**, 19, 2747-53	5.6	43
160	Conventional renal cancer in a patient with fumarate hydratase mutation. *Human Pathology*, **2007**, 38, 793-6	3.7	43
159	No evidence of somatic aryl hydrocarbon receptor interacting protein mutations in sporadic endocrine neoplasia. *Endocrine-Related Cancer*, **2007**, 14, 901-6	5.7	42
158	Aggressive pituitary adenomas occurring in young patients in a large Polynesian kindred with a germline R271W mutation in the AIP gene. *European Journal of Endocrinology*, **2009**, 161, 799-804	6.5	41
157	Carbonic anhydrase IX is highly expressed in hereditary nonpolyposis colorectal cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 1760-6	4	41
156	Impact of constitutional TET2 haploinsufficiency on molecular and clinical phenotype in humans. *Nature Communications*, **2019**, 10, 1252	17.4	40
155	Identification of candidate oncogenes in human colorectal cancers with microsatellite instability. *Gastroenterology*, **2013**, 145, 540-3.e22	13.3	40
154	Analysis of a Finnish family confirms RHBDF2 mutations as the underlying factor in tylosis with esophageal cancer. *Familial Cancer*, **2012**, 11, 525-8	3	40
153	Little evidence for involvement of MLH3 in colorectal cancer predisposition. *International Journal of Cancer*, **2003**, 106, 292-6	7.5	40
152	Common deletion of SMAD4 in juvenile polyposis is a mutational hotspot. *American Journal of Human Genetics*, **2002**, 70, 1357-62	11	40
151	Germline and somatic mutation analysis of MLH3 in MSI-positive colorectal cancer. *American Journal of Pathology*, **2000**, 157, 347-52	5.8	40
150	Frequent L1 retrotranspositions originating from TTC28 in colorectal cancer. *Oncotarget*, **2014**, 5, 853-9	3.3	40
149	High familial risk in nodular lymphocyte-predominant Hodgkin lymphoma. *Journal of Clinical Oncology*, **2013**, 31, 938-43	2.2	39
148	COGENT (COlorectal cancer GENeTics): an international consortium to study the role of polymorphic variation on the risk of colorectal cancer. *British Journal of Cancer*, **2010**, 102, 447-54	8.7	39

147	Mendelian randomisation analysis strongly implicates adiposity with risk of developing colorectal cancer. *British Journal of Cancer*, **2016**, 115, 266-72	8.7	39
146	Exome sequencing reveals three novel candidate predisposition genes for diffuse gastric cancer. *Familial Cancer*, **2015**, 14, 241-6	3	37
145	Intestinal cancer in patients with a germline mutation in the down-regulated in adenoma (DRA) gene. *Oncogene*, **1998**, 16, 681-4	9.2	37
144	Exome-wide somatic mutation characterization of small bowel adenocarcinoma. *PLoS Genetics*, **2018**, 14, e1007200	6	36
143	A Finnish founder mutation in RAD51D: analysis in breast, ovarian, prostate, and colorectal cancer. *Journal of Medical Genetics*, **2012**, 49, 429-32	5.8	35
142	High frequency of RPL22 mutations in microsatellite-unstable colorectal and endometrial tumors. *Human Mutation*, **2014**, 35, 1442-5	4.7	34
141	Complexity of 12q13-22 amplicon in liposarcoma: microsatellite repeat analysis. *Genes Chromosomes and Cancer*, **1997**, 18, 66-70	5	33
140	Background mutation frequency in microsatellite-unstable colorectal cancer. *Cancer Research*, **2007**, 67, 5691-8	10.1	33
139	CHEK2 1100delC and colorectal cancer. *Journal of Medical Genetics*, **2003**, 40, e110	5.8	33
138	No evidence of RET germline mutations in familial pituitary adenoma. *Journal of Molecular Endocrinology*, **2011**, 46, 1-8	4.5	32
137	Allelic analysis of serous ovarian carcinoma reveals two putative tumor suppressor loci at 18q22-q23 distal to SMAD4, SMAD2, and DCC. *American Journal of Pathology*, **2001**, 159, 35-42	5.8	32
136	NOD2 3020insC alone is not sufficient for colorectal cancer predisposition. *Cancer Research*, **2004**, 64, 7245-7	10.1	31
135	No SMAD4 hypermethylation in colorectal cancer. *British Journal of Cancer*, **2000**, 83, 1015-9	8.7	31
134	Modifiable pathways for colorectal cancer: a mendelian randomisation analysis. *The Lancet Gastroenterology and Hepatology*, **2020**, 5, 55-62	18.8	31
133	Oncogenic exon 2 mutations in Mediator subunit MED12 disrupt allosteric activation of cyclin C-CDK8/19. *Journal of Biological Chemistry*, **2018**, 293, 4870-4882	5.4	30
132	Exomic landscape of MED12 mutation-negative and -positive uterine leiomyomas. *International Journal of Cancer*, **2014**, 134, 1008-12	7.5	30
131	7q deletion mapping and expression profiling in uterine fibroids. *Oncogene*, **2005**, 24, 6545-54	9.2	30
130	Screening for microsatellite instability target genes in colorectal cancers. *Journal of Medical Genetics*, **2002**, 39, 785-9	5.8	30

129	MED12 mutations and FH inactivation are mutually exclusive in uterine leiomyomas. *British Journal of Cancer*, **2016**, 114, 1405-11	8.7	29
128	An A13 repeat within the 3Runtranslated region of epidermal growth factor receptor (EGFR) is frequently mutated in microsatellite instability colon cancers and is associated with increased EGFR expression. *Cancer Research*, **2009**, 69, 7811-8	10.1	29
127	Preferential amplification of AURKA 91A (Ile31) in familial colorectal cancers. *International Journal of Cancer*, **2006**, 118, 505-8	7.5	29
126	Functional diversity of human protection of telomeres 1 isoforms in telomere protection and cellular senescence. *Cancer Research*, **2007**, 67, 11677-86	10.1	29
125	Mutation analysis of three genes encoding novel LKB1-interacting proteins, BRG1, STRADalpha, and MO25alpha, in Peutz-Jeghers syndrome. *British Journal of Cancer*, **2005**, 92, 1126-9	8.7	28
124	Variation at 2q35 (PNKD and TMBIM1) influences colorectal cancer risk and identifies a pleiotropic effect with inflammatory bowel disease. *Human Molecular Genetics*, **2016**, 25, 2349-2359	5.6	27
123	Multiple clinical characteristics separate MED12-mutation-positive and -negative uterine leiomyomas. *Scientific Reports*, **2017**, 7, 1015	4.9	27
122	Variants in the netrin-1 receptor UNC5C prevent apoptosis and increase risk of familial colorectal cancer. *Gastroenterology*, **2011**, 141, 2039-46	13.3	27
121	Mutation screening of fumarate hydratase by multiplex ligation-dependent probe amplification: detection of exonic deletion in a patient with leiomyomatosis and renal cell cancer. *Cancer Genetics and Cytogenetics*, **2008**, 183, 83-8		27
120	Retrotransposon insertions can initiate colorectal cancer and are associated with poor survival. *Nature Communications*, **2019**, 10, 4022	17.4	26
119	COGENT (COlorectal cancer GENeTics) revisited. *Mutagenesis*, **2012**, 27, 143-51	2.8	26
118	Genetic predisposition to uterine leiomyoma is determined by loci for genitourinary development and genome stability. *ELife*, **2018**, 7,	8.9	26
117	Phosphoprotein Keratin 23 accumulates in MSS but not MSI colon cancers in vivo and impacts viability and proliferation in vitro. *Molecular Oncology*, **2007**, 1, 181-95	7.9	25
116	Colorectal pretumor progression before and after loss of DNA mismatch repair. *American Journal of Pathology*, **2004**, 164, 1447-53	5.8	25
115	The C/C-13910 genotype of adult-type hypolactasia is associated with an increased risk of colorectal cancer in the Finnish population. *Gut*, **2005**, 54, 643-7	19.2	25
114	CDX2 mutations do not account for juvenile polyposis or Peutz-Jeghers syndrome and occur infrequently in sporadic colorectal cancers. *British Journal of Cancer*, **2001**, 84, 1314-6	8.7	25
113	Germline mutations in young non-smoking women with lung adenocarcinoma. *Lung Cancer*, **2018**, 122, 76-82	5.9	25
112	Identification of 33 candidate oncogenes by screening for base-specific mutations. *British Journal of Cancer*, **2014**, 111, 1657-62	8.7	24

111	Downregulation of SRF-FOS-JUNB pathway in fumarate hydratase deficiency and in uterine leiomyomas. *Oncogene*, **2009**, 28, 1261-73	9.2	24
110	No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. *Familial Cancer*, **2010**, 9, 245-51	3	24
109	Somatic mutations and germline sequence variants in patients with familial colorectal cancer. *International Journal of Cancer*, **2010**, 127, 2974-80	7.5	24
108	Tumour selection advantage of non-dominant negative P53 mutations in homozygotic MDM2-SNP309 colorectal cancer cells. *Journal of Medical Genetics*, **2007**, 44, 75-80	5.8	24
107	Germline fumarate hydratase mutations in patients with ovarian mucinous cystadenoma. *European Journal of Human Genetics*, **2006**, 14, 880-3	5.3	24
106	Array comparative genomic hybridization identifies a distinct DNA copy number profile in renal cell cancer associated with hereditary leiomyomatosis and renal cell cancer. *Genes Chromosomes and Cancer*, **2009**, 48, 544-51	5	23
105	Strong family history of uterine leiomyomatosis warrants fumarate hydratase mutation screening. *Human Reproduction*, **2012**, 27, 1865-9	5.7	23
104	Hereditary intestinal cancer. *Seminars in Cancer Biology*, **2000**, 10, 289-98	12.7	23
103	Characterization of the 17p amplicon in human sarcomas: microsatellite marker analysis. *International Journal of Cancer*, **1999**, 82, 329-33	7.5	23
102	Susceptibility to pituitary neoplasia related to MEN-1, CDKN1B and AIP mutations: an update. *Human Molecular Genetics*, **2007**, 16 Spec No 1, R73-9	5.6	22
101	Tracing cell fates in human colorectal tumors from somatic microsatellite mutations: evidence of adenomas with stem cell architecture. *American Journal of Pathology*, **1998**, 153, 1189-200	5.8	22
100	Genome-wide association study and meta-analysis in Northern European populations replicate multiple colorectal cancer risk loci. *International Journal of Cancer*, **2018**, 142, 540-546	7.5	21
99	Villin expression is frequently lost in poorly differentiated colon cancer. *American Journal of Pathology*, **2012**, 180, 1509-21	5.8	21
98	Low-penetrance susceptibility variants in familial colorectal cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 1478-83	4	21
97	Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours. *Human Molecular Genetics*, **2010**, 19, 697-706	5.6	21
96	Somatic mutation analysis of MYH11 in breast and prostate cancer. *BMC Cancer*, **2008**, 8, 263	4.8	21
95	Germline deletions of EXO1 do not cause colorectal tumors and lesions which are null for EXO1 do not have microsatellite instability. *Cancer Genetics and Cytogenetics*, **2003**, 147, 121-7		21
94	E-cadherin is not frequently mutated in hereditary gastric cancer. *Journal of Medical Genetics*, **2001**, 38, 49-52	5.8	21

93	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget*, **2014**, 5, 8223-34	3.3	21
92	Systematic search for rare variants in Finnish early-onset colorectal cancer patients. *Cancer Genetics* , **2015**, 208, 35-40	2.3	20
91	Characterization of the colorectal cancer-associated enhancer MYC-335 at 8q24: the role of rs67491583. *Cancer Genetics*, **2012**, 205, 25-33	2.3	20
90	Cancer risk in mutation carriers of DNA-mismatch-repair genes **1999**, 81, 214		20
89	Increased HIF1 alpha in SDH and FH deficient tumors does not cause microsatellite instability. *International Journal of Cancer*, **2007**, 121, 1386-9	7.5	19
88	Whole-Genome Sequencing Identifies STAT4 as a Putative Susceptibility Gene in Classic Kaposi Sarcoma. *Journal of Infectious Diseases*, **2015**, 211, 1842-51	7	18
87	No evidence for association of NOD2 R702W and G908R with colorectal cancer. *International Journal of Cancer*, **2007**, 121, 76-9	7.5	18
86	A novel functionally deficient MYH variant in individuals with colorectal adenomatous polyposis. *Human Mutation*, **2005**, 26, 393	4.7	18
85	Discovery of potential causative mutations in human coding and noncoding genome with the interactive software BasePlayer. *Nature Protocols*, **2018**, 13, 2580-2600	18.8	18
84	Mutation analysis of components of the Mediator kinase module in MED12 mutation-negative uterine leiomyomas. *British Journal of Cancer*, **2014**, 110, 2246-9	8.7	17
83	Primary mediastinal large B-cell lymphoma segregating in a family: exome sequencing identifies MLL as a candidate predisposition gene. *Blood*, **2013**, 121, 3428-30	2.2	17
82	Refinement of the associations between risk of colorectal cancer and polymorphisms on chromosomes 1q41 and 12q13.13. *Human Molecular Genetics*, **2012**, 21, 934-46	5.6	17
81	Comprehensive Evaluation of Protein Coding Mononucleotide Microsatellites in Microsatellite-Unstable Colorectal Cancer. *Cancer Research*, **2017**, 77, 4078-4088	10.1	16
80	Systematic search for enhancer elements and somatic allelic imbalance at seven low-penetrance colorectal cancer predisposition loci. *BMC Medical Genetics*, **2011**, 12, 23	2.1	16
79	Smad4 haploinsufficiency: a matter of dosage. *PathoGenetics*, **2008**, 1, 2		16
78	No evidence for dual role of loss of heterozygosity in hereditary non-polyposis colorectal cancer. *Oncogene*, **2007**, 26, 2513-7	9.2	16
77	Global metabolomic profiling of uterine leiomyomas. *British Journal of Cancer*, **2017**, 117, 1855-1864	8.7	15
76	Identification of homozygous deletion in ACAN and other candidate variants in familial classical Hodgkin lymphoma by exome sequencing. *British Journal of Haematology*, **2015**, 170, 428-31	4.5	15

75	EPHB2 germline variants in patients with colorectal cancer or hyperplastic polyposis. *BMC Cancer*, **2006**, 6, 145	4.8	15
74	Mutations in two short noncoding mononucleotide repeats in most microsatellite-unstable colorectal cancers. *Cancer Research*, **2005**, 65, 4607-13	10.1	15
73	Genetic predisposition and somatic diversification in tumor development and progression. *Advances in Cancer Research*, **2001**, 80, 83-114	5.9	15
72	PPP2R1B gene in chronic lymphocytic leukemias and mantle cell lymphomas. *Leukemia and Lymphoma*, **2001**, 41, 177-83	1.9	15
71	Candidate susceptibility variants for esophageal squamous cell carcinoma. *Genes Chromosomes and Cancer*, **2017**, 56, 453-459	5	14
70	Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer. *PLoS Genetics*, **2020**, 16, e1008572	6	14
69	Detection of subclonal L1 transductions in colorectal cancer by long-distance inverse-PCR and Nanopore sequencing. *Scientific Reports*, **2017**, 7, 14521	4.9	14
68	Clonally related uterine leiomyomas are common and display branched tumor evolution. *Human Molecular Genetics*, **2015**, 24, 4407-16	5.6	14
67	Genome-wide allelotyping of 104 Finnish colorectal cancers reveals an excess of allelic imbalance in chromosome 20q in familial cases. *Oncogene*, **2003**, 22, 2206-14	9.2	14
66	No evidence of microsatellite instability in bone tumours. *British Journal of Cancer*, **1996**, 74, 453-5	8.7	14
65	Clinical characterization, genetic mapping and whole-genome sequence analysis of a novel autosomal recessive intellectual disability syndrome. *European Journal of Medical Genetics*, **2014**, 57, 543-51	2.6	13
64	PolyA deletions in hereditary nonpolyposis colorectal cancer: mutations before a gatekeeper. *American Journal of Pathology*, **2002**, 160, 1503-6	5.8	13
63	Multiple components of PKA and TGF-β pathways are mutated in pseudomyxoma peritonei. *PLoS ONE*, **2017**, 12, e0174898	3.7	13
62	Germline determinants of the somatic mutation landscape in 2,642 cancer genomes		13
61	Deficient H2A.Z deposition is associated with genesis of uterine leiomyoma. *Nature*, **2021**, 596, 398-403	50.4	13
60	Exome sequencing in diagnostic evaluation of colorectal cancer predisposition in young patients. *Scandinavian Journal of Gastroenterology*, **2013**, 48, 672-8	2.4	12
59	Numbers of mutations to different types of colorectal cancer. *BMC Cancer*, **2005**, 5, 126	4.8	12
58	No MSH6 germline mutations in breast cancer families with colorectal and/or endometrial cancer. *Journal of Medical Genetics*, **2005**, 42, e22	5.8	12

57	Chromothripsis in uterine leiomyomas. *New England Journal of Medicine*, **2013**, 369, 2160-1	59.2	11
56	High frequency of TTK mutations in microsatellite-unstable colorectal cancer and evaluation of their effect on spindle assembly checkpoint. *Carcinogenesis*, **2011**, 32, 305-11	4.6	11
55	Genetic and Epigenetic Characterization of Growth Hormone-Secreting Pituitary Tumors. *Molecular Cancer Research*, **2019**, 17, 2432-2443	6.6	11
54	Contribution of allelic imbalance to colorectal cancer. *Nature Communications*, **2018**, 9, 3664	17.4	11
53	Nationwide registry-based analysis of cancer clustering detects strong familial occurrence of Kaposi sarcoma. *PLoS ONE*, **2013**, 8, e55209	3.7	10
52	Genetic and Epigenetic Characteristics of Inflammatory Bowel Disease-Associated Colorectal Cancer. *Gastroenterology*, **2021**, 161, 592-607	13.3	10
51	Whole-exome sequencing identifies novel candidate predisposition genes for familial polycythemia vera. *Human Genomics*, **2017**, 11, 6	6.8	9
50	Microsatellite markers as tools for characterization of DNA amplifications evaluated by comparative genomic hybridization. *Cancer Genetics and Cytogenetics*, **1997**, 93, 33-8		9
49	Definition of a minimal region of deletion of chromosome 7 in uterine leiomyomas by tiling-path microarray CGH and mutation analysis of known genes in this region. *Genes Chromosomes and Cancer*, **2007**, 46, 451-8	5	9
48	A missense mutation in the BRCA2 gene in three siblings with ovarian cancer. *British Journal of Cancer*, **1998**, 77, 1199-202	8.7	9
47	New target genes in endometrial tumors show a role for the estrogen-receptor pathway in microsatellite-unstable cancers. *Human Mutation*, **2014**, 35, 1514-23	4.7	8
46	Mutation and LOH analysis of ACO2 in colorectal cancer: no evidence of biallelic genetic inactivation. *Journal of Medical Genetics*, **2003**, 40, e73	5.8	8
45	No fumarate hydratase (FH) mutations in hereditary prostate cancer. *Journal of Medical Genetics*, **2003**, 40, e19	5.8	8
44	Screening of Finnish RAD51C founder mutations in prostate and colorectal cancer patients. *BMC Cancer*, **2012**, 12, 552	4.8	7
43	Analysis of KLHDC8B in familial nodular lymphocyte predominant Hodgkin lymphoma. *British Journal of Haematology*, **2011**, 154, 413-5	4.5	7
42	ARLTS1 germline variants and the risk for breast, prostate, and colorectal cancer. *European Journal of Human Genetics*, **2008**, 16, 983-91	5.3	7
41	No germline FH mutations in familial breast cancer patients. *European Journal of Human Genetics*, **2005**, 13, 506-9	5.3	7
40	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). *Genetics in Medicine*, **2019**, 21, 2355-2363	8.1	6

39	Comprehensive evaluation of coding region point mutations in microsatellite-unstable colorectal cancer. *EMBO Molecular Medicine*, **2018**, 10,	12	6
38	Candidate susceptibility variants in angioimmunoblastic T-cell lymphoma. *Familial Cancer*, **2019**, 18, 113-119	3	6
37	Downregulation of the hedgehog receptor PTCH1 in colorectal serrated adenocarcinomas is not caused by PTCH1 mutations. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2011**, 458, 213-9	5.1	5
36	Blood-derived gene-expression profiling in unravelling susceptibility to recessive disease. *Journal of Medical Genetics*, **2007**, 44, 718-20	5.8	5
35	Colibactin DNA damage signature indicates causative role in colorectal cancer		5
34	Exome and immune cell score analyses reveal great variation within synchronous primary colorectal cancers. *British Journal of Cancer*, **2019**, 120, 922-930	8.7	4
33	Trilateral retinoblastoma in a patient with Peutz-Jeghers syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1096-100	2.5	4
32	Germline MSH6 Mutation in a Patient With Two Independent Primary Glioblastomas. *Journal of Neuropathology and Experimental Neurology*, **2017**, 76, 848-853	3.1	4
31	Modeling tumor predisposing FH mutations in yeast: effects on fumarase activity, growth phenotype and gene expression profile. *International Journal of Cancer*, **2006**, 118, 1340-5	7.5	4
30	Aryl hydrocarbon receptor-interacting protein and acromegaly. *Hormone Research in Paediatrics*, **2007**, 68 Suppl 5, 127-31	3.3	4
29	Role of TP53 P72R polymorphism in human papillomavirus associated premalignant laryngeal neoplasm. *Journal of Medical Genetics*, **2001**, 38, 327	5.8	4
28	MSH2 and MLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis **1997**, 18, 269		4
27	A 17p11.2 germline deletion in a patient with Smith-Magenis syndrome and neuroblastoma. *Journal of Medical Genetics*, **2005**, 42, e3	5.8	3
26	BasePlayer: Versatile Analysis Software for Large-scale Genomic Variant Discovery		3
25	No evidence of EMAST in whole genome sequencing data from 248 colorectal cancers. *Genes Chromosomes and Cancer*, **2021**, 60, 463-473	5	3
24	Cancer risk in mutation carriers of DNA-mismatch-repair genes **1999**, 81, 214		3
23	3RUTR poly(T/U) repeat of EWSR1 is altered in microsatellite unstable colorectal cancer with nearly perfect sensitivity. *Familial Cancer*, **2015**, 14, 449-53	3	2
22	Computational identification of candidate loci for recessively inherited mutation using high-throughput SNP arrays. *Bioinformatics*, **2007**, 23, 1952-61	7.2	2

21	Mutation analysis of SMAD2, SMAD3, and SMAD4 genes in hereditary non-polyposis colorectal. *Journal of Medical Genetics*, **2000**, 37, 298-300	5.8	2
20	Prognostic Value of Immune Environment Analysis in Small Bowel Adenocarcinomas with Verified Mutational Landscape and Predisposing Conditions. *Cancers*, **2020**, 12,	6.6	2
19	From APC to the genetics of hereditary and familial colon cancer syndromes. *Human Molecular Genetics*, **2021**, 30, R206-R224	5.6	2
18	Candidate Predisposition Variants in Kaposi Sarcoma as Detected by Whole-Genome Sequencing. *Open Forum Infectious Diseases*, **2019**, 6, ofz337	1	1
17	Mutation analysis of MYH11 in acute myeloid leukemia. *Leukemia and Lymphoma*, **2008**, 49, 1829-31	1.9	1
16	Stress-induced expression of a novel variant of human fumarate hydratase (FH). *Gene Expression*, **2007**, 14, 59-69	3.4	1
15	Defining eligible patients for allele-selective chemotherapies targeting NAT2 in colorectal cancer. *Scientific Reports*, **2020**, 10, 22436	4.9	1
14	Identification of ZBTB18 as a novel colorectal tumor suppressor gene through genome-wide promoter hypermethylation analysis. *Clinical Epigenetics*, **2021**, 13, 88	7.7	1
13	Lack of an association between gallstone disease and bilirubin levels with risk of colorectal cancer: a Mendelian randomisation analysis. *British Journal of Cancer*, **2021**, 124, 1169-1174	8.7	1
12	Next-generation sequencing in a large pedigree segregating visceral artery aneurysms suggests potential role of COL4A1/COL4A2 in disease etiology. *Vascular*, **2021**, 17085381211033157	1.3	1
11	WNT2 activation through proximal germline deletion predisposes to small intestinal neuroendocrine tumors and intestinal adenocarcinomas. *Human Molecular Genetics*, **2021**, 30, 2429-2440	5.6	1
10	Human cell transformation by combined lineage conversion and oncogene expression. *Oncogene*, **2021**, 40, 5533-5547	9.2	1
9	Parity associates with chromosomal damage in uterine leiomyomas. *Nature Communications*, **2021**, 12, 5448	17.4	1
8	Uterine leiomyomas in hereditary leiomyomatosis and renal cell cancer (HLRCC) syndrome can be identified through distinct clinical characteristics and typical morphology. *Acta Obstetricia Et Gynecologica Scandinavica*, **2021**, 100, 2066-2075	3.8	0
7	Rule-based induction method for haplotype comparison and identification of candidate disease loci. *Genome Medicine*, **2012**, 4, 21	14.4	
6	Reply to the Letter to the Editor by Watanabe et al.. *Clinical Cancer Research*, **2006**, 12, 1654.1-1655	12.9	
5	17 Direct Sequencing for Peutz-Jeghers Gene LKB1 (STK11) Mutations. *Methods in Molecular Medicine*, **2001**, 50, 175-83		
4	Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572		

List of Publications

3 Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572

2 Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572

1 Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572