Lauri A Aaltonen

List of Publications by Year in Descending Order

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	Identification of ZBTB18 as a novel colorectal tumor suppressor gene through genome-wide promoter hypermethylation analysis. *Clinical Epigenetics*, **2021**, 13, 88	7.7	1
307	From APC to the genetics of hereditary and familial colon cancer syndromes. *Human Molecular Genetics*, **2021**, 30, R206-R224	5.6	2
306	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
305	No evidence of EMAST in whole genome sequencing data from 248 colorectal cancers. *Genes Chromosomes and Cancer*, **2021**, 60, 463-473	5	3
304	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
303	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
302	Human cell transformation by combined lineage conversion and oncogene expression. *Oncogene*, **2021**, 40, 5533-5547	9.2	1
301	Genetic and Epigenetic Characteristics of Inflammatory Bowel Disease-Associated Colorectal Cancer. *Gastroenterology*, **2021**, 161, 592-607	13.3	10
300	Deficient H2A.Z deposition is associated with genesis of uterine leiomyoma. *Nature*, **2021**, 596, 398-403	50.4	13
299	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
298	Parity associates with chromosomal damage in uterine leiomyomas. *Nature Communications*, **2021**, 12, 5448	17.4	1
297	Colibactin DNA-damage signature indicates mutational impact in colorectal cancer. *Nature Medicine* , **2020**, 26, 1063-1069	50.5	76
296	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
295	Defining eligible patients for allele-selective chemotherapies targeting NAT2 in colorectal cancer. *Scientific Reports*, **2020**, 10, 22436	4.9	1
294	Modifiable pathways for colorectal cancer: a mendelian randomisation analysis. *The Lancet Gastroenterology and Hepatology*, **2020**, 5, 55-62	18.8	31
293	Prognostic Value of Immune Environment Analysis in Small Bowel Adenocarcinomas with Verified Mutational Landscape and Predisposing Conditions. *Cancers*, **2020**, 12,	6.6	2
292	Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572		

291	Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572		
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289	Mutational processes of distinct POLE exonuclease domain mutants drive an enrichment of a specific TP53 mutation in colorectal cancer **2020**, 16, e1008572		
288	Candidate Predisposition Variants in Kaposi Sarcoma as Detected by Whole-Genome Sequencing. *Open Forum Infectious Diseases*, **2019**, 6, ofz337	1	1
287	Retrotransposon insertions can initiate colorectal cancer and are associated with poor survival. *Nature Communications*, **2019**, 10, 4022	17.4	26
286	Association analyses identify 31 new risk loci for colorectal cancer susceptibility. *Nature Communications*, **2019**, 10, 2154	17.4	81
285	Impact of constitutional TET2 haploinsufficiency on molecular and clinical phenotype in humans. *Nature Communications*, **2019**, 10, 1252	17.4	40
284	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
283	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
282	Candidate susceptibility variants in angioimmunoblastic T-cell lymphoma. *Familial Cancer*, **2019**, 18, 113-119	3	6
281	Genetic and Epigenetic Characterization of Growth Hormone-Secreting Pituitary Tumors. *Molecular Cancer Research*, **2019**, 17, 2432-2443	6.6	11
280	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
279	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
278	Comprehensive evaluation of coding region point mutations in microsatellite-unstable colorectal cancer. *EMBO Molecular Medicine*, **2018**, 10,	12	6
277	Exome-wide somatic mutation characterization of small bowel adenocarcinoma. *PLoS Genetics*, **2018**, 14, e1007200	6	36
276	Genetic predisposition to uterine leiomyoma is determined by loci for genitourinary development and genome stability. *ELife*, **2018**, 7,	8.9	26
275	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
274	Contribution of allelic imbalance to colorectal cancer. *Nature Communications*, **2018**, 9, 3664	17.4	11

273	Germline mutations in young non-smoking women with lung adenocarcinoma. *Lung Cancer*, **2018**, 122, 76-82	5.9	25
272	Candidate susceptibility variants for esophageal squamous cell carcinoma. *Genes Chromosomes and Cancer*, **2017**, 56, 453-459	5	14
271	Comprehensive Evaluation of Protein Coding Mononucleotide Microsatellites in Microsatellite-Unstable Colorectal Cancer. *Cancer Research*, **2017**, 77, 4078-4088	10.1	16
270	Mendelian randomisation implicates hyperlipidaemia as a risk factor for colorectal cancer. *International Journal of Cancer*, **2017**, 140, 2701-2708	7.5	50
269	Global metabolomic profiling of uterine leiomyomas. *British Journal of Cancer*, **2017**, 117, 1855-1864	8.7	15
268	Pro-inflammatory fatty acid profile and colorectal cancer risk: A Mendelian randomisation analysis. *European Journal of Cancer*, **2017**, 84, 228-238	7.5	56
267	Multiple clinical characteristics separate MED12-mutation-positive and -negative uterine leiomyomas. *Scientific Reports*, **2017**, 7, 1015	4.9	27
266	Germline MSH6 Mutation in a Patient With Two Independent Primary Glioblastomas. *Journal of Neuropathology and Experimental Neurology*, **2017**, 76, 848-853	3.1	4
265	Detection of subclonal L1 transductions in colorectal cancer by long-distance inverse-PCR and Nanopore sequencing. *Scientific Reports*, **2017**, 7, 14521	4.9	14
264	Whole-exome sequencing identifies novel candidate predisposition genes for familial polycythemia vera. *Human Genomics*, **2017**, 11, 6	6.8	9
263	Multiple components of PKA and TGF-pathways are mutated in pseudomyxoma peritonei. *PLoS ONE*, **2017**, 12, e0174898	3.7	13
262	MED12 mutations and FH inactivation are mutually exclusive in uterine leiomyomas. *British Journal of Cancer*, **2016**, 114, 1405-11	8.7	29
261	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
260	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
259	Mendelian randomisation analysis strongly implicates adiposity with risk of developing colorectal cancer. *British Journal of Cancer*, **2016**, 115, 266-72	8.7	39
258	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
257	Systematic search for rare variants in Finnish early-onset colorectal cancer patients. *Cancer Genetics* , **2015**, 208, 35-40	2.3	20
256	Whole-Genome Sequencing of Growth Hormone (GH)-Secreting Pituitary Adenomas. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 3918-27	5.6	62

255	Whole-Genome Sequencing Identifies STAT4 as a Putative Susceptibility Gene in Classic Kaposi Sarcoma. *Journal of Infectious Diseases*, **2015**, 211, 1842-51	7	18
254	Genomic profile of pseudomyxoma peritonei analyzed using next-generation sequencing and immunohistochemistry. *International Journal of Cancer*, **2015**, 136, E282-9	7.5	52
253	CTCF/cohesin-binding sites are frequently mutated in cancer. *Nature Genetics*, **2015**, 47, 818-21	36.3	286
252	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
251	Clonally related uterine leiomyomas are common and display branched tumor evolution. *Human Molecular Genetics*, **2015**, 24, 4407-16	5.6	14
250	Exome sequencing reveals three novel candidate predisposition genes for diffuse gastric cancer. *Familial Cancer*, **2015**, 14, 241-6	3	37
249	High frequency of RPL22 mutations in microsatellite-unstable colorectal and endometrial tumors. *Human Mutation*, **2014**, 35, 1442-5	4.7	34
248	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
247	Genomics of uterine leiomyomas: insights from high-throughput sequencing. *Fertility and Sterility*, **2014**, 102, 621-9	4.8	112
246	MED12 mutation frequency in unselected sporadic uterine leiomyomas. *Fertility and Sterility*, **2014**, 102, 1137-42	4.8	51
245	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
244	Identification of 33 candidate oncogenes by screening for base-specific mutations. *British Journal of Cancer*, **2014**, 111, 1657-62	8.7	24
243	Uterine leiomyoma-linked MED12 mutations disrupt mediator-associated CDK activity. *Cell Reports*, **2014**, 7, 654-60	10.6	99
242	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
241	Exomic landscape of MED12 mutation-negative and -positive uterine leiomyomas. *International Journal of Cancer*, **2014**, 134, 1008-12	7.5	30
240	Frequent L1 retrotranspositions originating from TTC28 in colorectal cancer. *Oncotarget*, **2014**, 5, 853-9	3.3	40
239	Population distribution and ancestry of the cancer protective MDM2 SNP285 (rs117039649). *Oncotarget*, **2014**, 5, 8223-34	3.3	21
238	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

237	Characterization of uterine leiomyomas by whole-genome sequencing. *New England Journal of Medicine*, **2013**, 369, 43-53	59.2	223
236	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
235	Diagnostic cancer genome sequencing and the contribution of germline variants. *Science*, **2013**, 339, 1559-62	33.3	49
234	Identification of candidate oncogenes in human colorectal cancers with microsatellite instability. *Gastroenterology*, **2013**, 145, 540-3.e22	13.3	40
233	High familial risk in nodular lymphocyte-predominant Hodgkin lymphoma. *Journal of Clinical Oncology*, **2013**, 31, 938-43	2.2	39
232	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
231	MED12 exon 2 mutations in histopathological uterine leiomyoma variants. *European Journal of Human Genetics*, **2013**, 21, 1300-3	5.3	55
230	Trilateral retinoblastoma in a patient with Peutz-Jeghers syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1096-100	2.5	4
229	Exome sequencing in diagnostic evaluation of colorectal cancer predisposition in young patients. *Scandinavian Journal of Gastroenterology*, **2013**, 48, 672-8	2.4	12
228	Chromothripsis in uterine leiomyomas. *New England Journal of Medicine*, **2013**, 369, 2160-1	59.2	11
227	Lessons from functional analysis of genome-wide association studies. *Cancer Research*, **2013**, 73, 4180-4	10.1	50
226	Eleven candidate susceptibility genes for common familial colorectal cancer. *PLoS Genetics*, **2013**, 9, e1003876		57
225	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
224	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
223	Nationwide registry-based analysis of cancer clustering detects strong familial occurrence of Kaposi sarcoma. *PLoS ONE*, **2013**, 8, e55209	3.7	10
222	Candidate driver genes in microsatellite-unstable colorectal cancer. *International Journal of Cancer*, **2012**, 130, 1558-66	7.5	78
221	Common variation near CDKN1A, POLD3 and SHROOM2 influences colorectal cancer risk. *Nature Genetics*, **2012**, 44, 770-6	36.3	184
220	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

219	Villin expression is frequently lost in poorly differentiated colon cancer. *American Journal of Pathology*, **2012**, 180, 1509-21	5.8	21
218	Characterization of the colorectal cancer-associated enhancer MYC-335 at 8q24: the role of rs67491583. *Cancer Genetics*, **2012**, 205, 25-33	2.3	20
217	Identification of Lynch syndrome among patients with colorectal cancer. *JAMA - Journal of the American Medical Association*, **2012**, 308, 1555-65	27.4	323
216	Screening of Finnish RAD51C founder mutations in prostate and colorectal cancer patients. *BMC Cancer*, **2012**, 12, 552	4.8	7
215	Somatic MED12 mutations in uterine leiomyosarcoma and colorectal cancer. *British Journal of Cancer*, **2012**, 107, 1761-5	8.7	91
214	Segregation of a missense variant in enteric smooth muscle actin ￼2 with autosomal dominant familial visceral myopathy. *Gastroenterology*, **2012**, 143, 1482-1491.e3	13.3	71
213	Loss of SUFU function in familial multiple meningioma. *American Journal of Human Genetics*, **2012**, 91, 520-6	11	103
212	Mice lacking a Myc enhancer that includes human SNP rs6983267 are resistant to intestinal tumors. *Science*, **2012**, 338, 1360-3	33.3	171
211	Rule-based induction method for haplotype comparison and identification of candidate disease loci. *Genome Medicine*, **2012**, 4, 21	14.4	
210	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
209	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
208	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
207	Strong family history of uterine leiomyomatosis warrants fumarate hydratase mutation screening. *Human Reproduction*, **2012**, 27, 1865-9	5.7	23
206	COGENT (COlorectal cancer GENeTics) revisited. *Mutagenesis*, **2012**, 27, 143-51	2.8	26
205	Variants in the netrin-1 receptor UNC5C prevent apoptosis and increase risk of familial colorectal cancer. *Gastroenterology*, **2011**, 141, 2039-46	13.3	27
204	Mutations in BRIP1 confer high risk of ovarian cancer. *Nature Genetics*, **2011**, 43, 1104-7	36.3	285
203	MED12, the mediator complex subunit 12 gene, is mutated at high frequency in uterine leiomyomas. *Science*, **2011**, 334, 252-5	33.3	449
202	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

201	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
200	Exome sequencing reveals germline NPAT mutation as a candidate risk factor for Hodgkin lymphoma. *Blood*, **2011**, 118, 493-8	2.2	65
199	Analysis of KLHDC8B in familial nodular lymphocyte predominant Hodgkin lymphoma. *British Journal of Haematology*, **2011**, 154, 413-5	4.5	7
198	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
197	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
196	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
195	No evidence of RET germline mutations in familial pituitary adenoma. *Journal of Molecular Endocrinology*, **2011**, 46, 1-8	4.5	32
194	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
193	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
192	MED12 exon 2 mutations are common in uterine leiomyomas from South African patients. *Oncotarget*, **2011**, 2, 966-9	3.3	78
191	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
190	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
189	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
188	Mutations in the circadian gene CLOCK in colorectal cancer. *Molecular Cancer Research*, **2010**, 8, 952-60	6.6	60
187	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
186	Low-penetrance susceptibility variants in familial colorectal cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2010**, 19, 1478-83	4	21
185	Recessively inherited right atrial isomerism caused by mutations in growth/differentiation factor 1 (GDF1). *Human Molecular Genetics*, **2010**, 19, 2747-53	5.6	43
184	Mixed lineage kinase 3 gene mutations in mismatch repair deficient gastrointestinal tumours. *Human Molecular Genetics*, **2010**, 19, 697-706	5.6	21

183	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
182	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
181	No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. *Familial Cancer*, **2010**, 9, 245-51	3	24
180	Somatic mutations and germline sequence variants in patients with familial colorectal cancer. *International Journal of Cancer*, **2010**, 127, 2974-80	7.5	24
179	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
178	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
177	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
176	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
175	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
174	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
173	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
172	A TARBP2 mutation in human cancer impairs microRNA processing and DICER1 function. *Nature Genetics*, **2009**, 41, 365-70	36.3	317
171	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
170	Downregulation of SRF-FOS-JUNB pathway in fumarate hydratase deficiency and in uterine leiomyomas. *Oncogene*, **2009**, 28, 1261-73	9.2	24
169	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
168	ARLTS1 germline variants and the risk for breast, prostate, and colorectal cancer. *European Journal of Human Genetics*, **2008**, 16, 983-91	5.3	7
167	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
166	Meta-analysis of genome-wide association data identifies four new susceptibility loci for colorectal cancer. *Nature Genetics*, **2008**, 40, 1426-35	36.3	457

165	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
164	Somatic mutation analysis of MYH11 in breast and prostate cancer. *BMC Cancer*, **2008**, 8, 263	4.8	21
163	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
162	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
161	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
160	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
159	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
158	Large genomic deletions in AIP in pituitary adenoma predisposition. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 4146-51	5.6	66
157	Mutation analysis of MYH11 in acute myeloid leukemia. *Leukemia and Lymphoma*, **2008**, 49, 1829-31	1.9	1
156	Allelic imbalance at rs6983267 suggests selection of the risk allele in somatic colorectal tumor evolution. *Cancer Research*, **2008**, 68, 14-7	10.1	64
155	Smad4 haploinsufficiency: a matter of dosage. *PathoGenetics*, **2008**, 1, 2		16
154	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
153	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
152	Development of colorectal tumors in colonoscopic surveillance in Lynch syndrome. *Gastroenterology*, **2007**, 133, 1093-8	13.3	110
151	No evidence for association of NOD2 R702W and G908R with colorectal cancer. *International Journal of Cancer*, **2007**, 121, 76-9	7.5	18
150	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
149	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
148	Serrated carcinomas form a subclass of colorectal cancer with distinct molecular basis. *Oncogene*, **2007**, 26, 312-20	9.2	118

147	No evidence for dual role of loss of heterozygosity in hereditary non-polyposis colorectal cancer. *Oncogene*, **2007**, 26, 2513-7	9.2	16
146	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
145	A recurrent mutation in PALB2 in Finnish cancer families. *Nature*, **2007**, 446, 316-9	50.4	349
144	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
143	Blood-derived gene-expression profiling in unravelling susceptibility to recessive disease. *Journal of Medical Genetics*, **2007**, 44, 718-20	5.8	5
142	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
141	Carbonic anhydrase IX is highly expressed in hereditary nonpolyposis colorectal cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2007**, 16, 1760-6	4	41
140	Differential expression of DHHC9 in microsatellite stable and instable human colorectal cancer subgroups. *British Journal of Cancer*, **2007**, 96, 1896-903	8.7	52
139	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
138	Computational identification of candidate loci for recessively inherited mutation using high-throughput SNP arrays. *Bioinformatics*, **2007**, 23, 1952-61	7.2	2
137	Germline CDKN1B/p27Kip1 mutation in multiple endocrine neoplasia. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 3321-5	5.6	230
136	No evidence of somatic aryl hydrocarbon receptor interacting protein mutations in sporadic endocrine neoplasia. *Endocrine-Related Cancer*, **2007**, 14, 901-6	5.7	42
135	Background mutation frequency in microsatellite-unstable colorectal cancer. *Cancer Research*, **2007** , 67, 5691-8	10.1	33
134	Clusterin expression in normal mucosa and colorectal cancer. *Molecular and Cellular Proteomics*, **2007**, 6, 1039-48	7.6	51
133	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
132	Functional diversity of human protection of telomeres 1 isoforms in telomere protection and cellular senescence. *Cancer Research*, **2007**, 67, 11677-86	10.1	29
131	Aryl hydrocarbon receptor-interacting protein and acromegaly. *Hormone Research in Paediatrics*, **2007**, 68 Suppl 5, 127-31	3.3	4
130	Stress-induced expression of a novel variant of human fumarate hydratase (FH). *Gene Expression*, **2007**, 14, 59-69	3.4	1

129	Conventional renal cancer in a patient with fumarate hydratase mutation. *Human Pathology*, **2007**, 38, 793-6	3.7	43
128	EPHB2 germline variants in patients with colorectal cancer or hyperplastic polyposis. *BMC Cancer*, **2006**, 6, 145	4.8	15
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List of Publications

3	BasePlayer: Versatile Analysis Software for Large-scale Genomic Variant Discovery	3
2	Germline determinants of the somatic mutation landscape in 2,642 cancer genomes	13
1	Colibactin DNA damage signature indicates causative role in colorectal cancer	5