Albert de la Chapelle

List of Publications by Year
in descending order

586	78,756	668 122	549 264
papers	citations	h-index	g-index

596	596	596	49606
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	PD-1 Blockade in Tumors with Mismatch-Repair Deficiency. New England Journal of Medicine, 2015, 372, 2509-2520.	13.9	7,696
2	Clues to the pathogenesis of familial colorectal cancer. Science, 1993, 260, 812-816.	6.0	2,563
3	Mutations of a mutS homolog in hereditary nonpolyposis colorectal cancer. Cell, 1993, 75, 1215-1225.	13.5	2,195
4	Hereditary Colorectal Cancer. New England Journal of Medicine, 2003, 348, 919-932.	13.9	1,870
5	Mutation of a mutL homolog in hereditary colon cancer. Science, 1994, 263, 1625-1629.	6.0	1,821
6	Mutations of two P/WS homologues in hereditary nonpolyposis colon cancer. Nature, 1994, 371, 75-80.	13.7	1,523
7	A serine/threonine kinase gene defective in Peutzâ€“Jeghers syndrome. Nature, 1998, 391, 184-187.	13.7	1,451
8	Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. Gastroenterology, 2000, 118, 829-834.	0.6	1,259
9	Screening for the Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer). New England Journal of Medicine, 2005, 352, 1851-1860.	13.9	1,237
10	Diverse spermatogenic defects in humans caused by Y chromosome deletions encompassing a novel RNAâ€“binding protein gene. Nature Genetics, 1995, 10, 383-393.	9.4	1,183
11	The role of microRNA genes in papillary thyroid carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 19075-19080.	3.3	1,137
12	Cancer risk in mutation carriers of DNA-mismatch-repair genes. , 1999, 81, 214-218.		1,061
13	Incidence of Hereditary Nonpolyposis Colorectal Cancer and the Feasibility of Molecular Screening for the Disease. New England Journal of Medicine, 1998, 338, 1481-1487.	13.9	1,048
14	Hypermutability and mismatch repair deficiency in RER+ tumor cells. Cell, 1993, 75, 1227-1236.	13.5	1,031
15	Mutation in the follicle-stimulating hormone receptor gene causes hereditary hypergonadotropic ovarian failure. Cell, 1995, 82, 959-968.	13.5	901
16	Analysis of mismatch repair genes in hereditary nonâ€“polyposis colorectal cancer patients. Nature Medicine, 1996, 2, 169-174.	15.2	892
17	The sex-determining region of the human Y chromosome encodes a finger protein. Cell, 1987, 51, 1091-1104.	13.5	881
18	Genetic mapping of a locus predisposing to human colorectal cancer. Science, 1993, 260, 810-812.	6.0	846

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19	Common SNP in pre-miR-146a decreases mature miR expression and predisposes to papillary thyroid carcinoma. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 7269-7274.	3.3	792
20	Feasibility of Screening for Lynch Syndrome Among Patients With Colorectal Cancer. Journal of Clinical Oncology, 2008, 26, 5783-5788.	0.8	760
21	Mismatch repair gene defects in sporadic colorectal cancers with microsatellite instability. Nature Genetics, 1995, 9, 48-55.	9.4	759
22	The diastrophic dysplasia gene encodes a novel sulfate transporter: Positional cloning by fine-structure linkage disequilibrium mapping. Cell, 1994, 78, 1073-1087.	13.5	731
23	Xâ€“linked anhidrotic (hypohidrotic) ectodermal dysplasia is caused by mutation in a novel transmembrane protein. Nature Genetics, 1996, 13, 409-416.	9.4	691
24	Mutations in the Gene Encoding Cystatin B in Progressive Myoclonus Epilepsy (EPM1). Science, 1996, 271, 1731-1734.	6.0	588
25	Genetic predisposition to colorectal cancer. Nature Reviews Cancer, 2004, 4, 769-780.	12.8	565
26	Screening for Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer) among Endometrial Cancer Patients. Cancer Research, 2006, 66, 7810-7817.	0.4	564
27	Genetic susceptibility to non-polyposis colorectal cancer. Journal of Medical Genetics, 1999, 36, 801-18.	1.5	549
28	Linkage disequilibrium mapping in isolated founder populations: diastrophic dysplasia in Finland. Nature Genetics, 1992, 2, 204-211.	9.4	544
29	Prevalence and Spectrum of Germline Cancer Susceptibility Gene Mutations Among Patients With Early-Onset Colorectal Cancer. JAMA Oncology, 2017, 3, 464.	3.4	510
30	Loss-of-Function Mutations in PPARÎ³ Associated with Human Colon Cancer. Molecular Cell, 1999, 3, 799-804.	4.5	485
31	The Clinical Phenotype of Lynch Syndrome Due to Germ-Line PMS2 Mutations. Gastroenterology, 2008, 135, 419-428.e1.	0.6	480
32	Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. Nature, 1986, 322, 73-77.	13.7	475
33	Population-Based Molecular Detection of Hereditary Nonpolyposis Colorectal Cancer. Journal of Clinical Oncology, 2000, 18, 2193-2200.	0.8	466
34	Mutations in the RNA Component of RNase MRP Cause a Pleiotropic Human Disease, Cartilage-Hair Hypoplasia. Cell, 2001, 104, 195-203.	13.5	461
35	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.	9.4	444
36	Identification of Lynch Syndrome Among Patients With Colorectal Cancer. JAMA - Journal of the American Medical Association, 2012, 308, 1555.	3.8	443

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37	Gene expression in papillary thyroid carcinoma reveals highly consistent profiles. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 15044-15049.	3.3	399
38	Mutations of the Downâ€“regulated in adenoma (DRA) gene cause congenital chloride diarrhoea. Nature Genetics, 1996, 14, 316-319.	9.4	394
39	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
40	Truncated erythropoietin receptor causes dominantly inherited benign human erythrocytosis.. Proceedings of the National Academy of Sciences of the United States of America, 1993, 90, 4495-4499.	3.3	370
41	A deletion in chromosome 22 can cause digeorge syndrome. Human Genetics, 1981, 57, 253-256.	1.8	363
42	Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. Nature Genetics, 2009, 41, 460-464.	9.4	353
43	Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer Syndrome: Later Age of Onset. Gastroenterology, 2005, 129, 415-421.	0.6	338
44	Downregulation of Death-Associated Protein Kinase 1 (DAPK1) in Chronic Lymphocytic Leukemia. Cell, 2007, 129, 879-890.	13.5	338
45	Reprogramming of miRNA networks in cancer and leukemia. Genome Research, 2010, 20, 589-599.	2.4	331
46	Risks of Lynch Syndrome Cancers for MSH6 Mutation Carriers. Journal of the National Cancer Institute, 2010, 102, 193-201.	3.0	328
47	Colon and Endometrial Cancers With Mismatch Repair Deficiency Can Arise From Somatic, Rather Than Germline, Mutations. Gastroenterology, 2014, 147, 1308-1316.e1.	0.6	328
48	Cohen Syndrome Is Caused by Mutations in a Novel Gene, COH1, Encoding a Transmembrane Protein with a Presumed Role in Vesicle-Mediated Sorting and Intracellular Protein Transport. American Journal of Human Genetics, 2003, 72, 1359-1369.	2.6	321
49	Polymorphic mature microRNAs from passenger strand of pre-miR-146a contribute to thyroid cancer. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 1502-1505.	3.3	311
50	Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer Syndrome: Later Age of Onset. Gastroenterology, 2005, 129, 415-421.	0.6	309
51	Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. Nature Genetics, 1994, 8, 405-410.	9.4	304
52	Mutations in the nebulin gene associated with autosomal recessive nemaline myopathy. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 2305-2310.	3.3	304
53	Disease gene mapping in isolated human populations: the example of Finland.. Journal of Medical Genetics, 1993, 30, 857-865.	1.5	302
54	Cubilin dysfunction causes abnormal metabolism of the steroid hormone 25(OH) vitamin D3. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 13895-13900.	3.3	288

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55	Gene expression and functional evidence of epithelial-to-mesenchymal transition in papillary thyroid carcinoma invasion. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 2803-2808.	3.3	285
56	hMSH2 mutations in hereditary nonpolyposis colorectal cancer kindreds. Cancer Research, 1994, 54, 4590-4.	0.4	280
57	The neuronal ceroid lipofuscinoses in human EPMR and mnd mutant mice are associated with mutations in CLN8. Nature Genetics, 1999, 23, 233-236.	9.4	277
58	Founding mutations and Alu-mediated recombination in hereditary colon cancer. Nature Medicine, 1995, 1, 1203-1206.	15.2	275
59	Clinical Relevance of Microsatellite Instability in Colorectal Cancer. Journal of Clinical Oncology, 2010, 28, 3380-3387.	0.8	273
60	A deletion map of the human Y chromosome based on DNA hybridization. American Journal of Human Genetics, 1986, 38, 109-24.	2.6	272
61	Expression profiling reveals fundamental biological differences in acute myeloid leukemia with isolated trisomy 8 and normal cytogenetics. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 1124-1129.	3.3	266
62	Galectin-3, fibronectin-1, CITED-1, HBME1 and cytokeratin-19 immunohistochemistry is useful for the differential diagnosis of thyroid tumors. Modern Pathology, 2005, 18, 48-57.	2.9	262
63	The functional cobalamin (vitamin B12)â€“intrinsic factor receptor is a novel complex of cubilin and amnionless. Blood, 2004, 103, 1573-1579.	0.6	259
64	Genetic and Epigenetic Modification of MLH1 Accounts for a Major Share of Microsatellite-Unstable Colorectal Cancers. American Journal of Pathology, 2000, 156, 1773-1779.	1.9	255
65	The intrinsic factorâ€“vitamin B12 receptor, cubilin, is a high-affinity apolipoprotein A-I receptor facilitating endocytosis of high-density lipoprotein. Nature Medicine, 1999, 5, 656-661.	15.2	248
66	Conversion of diploidy to haploidy. Nature, 2000, 403, 723-724.	13.7	248
67	Clinical significance of cytogenetics in acute myeloid leukemia. Seminars in Oncology, 1997, 24, 17-31.	0.8	247
68	The polymorphism rs944289 predisposes to papillary thyroid carcinoma through a large intergenic noncoding RNA gene of tumor suppressor type. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 8646-8651.	3.3	237
69	Mutations in CUBN, encoding the intrinsic factor-vitamin B12 receptor, cubilin, cause hereditary megaloblastic anaemia 1. Nature Genetics, 1999, 21, 309-313.	9.4	235
70	Assignment of an Usher syndrome type III (USH3) gene to chromosome 3q. Human Molecular Genetics, 1995, 4, 93-98.	1.4	227
71	Overexpression of the ETS-Related Gene, ERG, Predicts a Worse Outcome in Acute Myeloid Leukemia With Normal Karyotype: A Cancer and Leukemia Group B Study. Journal of Clinical Oncology, 2005, 23, 9234-9242.	0.8	226
72	The etiology of maleness in XX men. Human Genetics, 1981, 58, 105-116.	1.8	223

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73	Mutations in U4atac snRNA, a Component of the Minor Spliceosome, in the Developmental Disorder MOPD I. Science, 2011, 332, 238-240.	6.0	223
74	BAALC expression predicts clinical outcome of de novo acute myeloid leukemia patients with normal cytogenetics: a Cancer and Leukemia Group B Study. Blood, 2003, 102, 1613-1618.	0.6	222
75	Linkage disequilibrium mapping in isolated populations: The example of Finland revisited. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 12416-12423.	3.3	214
76	Pseudoautosomal DNA sequences in the pairing region of the human sex chromosomes. Nature, 1985, 317, 692-697.	13.7	212
77	Discovery of common variants associated with low TSH levels and thyroid cancer risk. Nature Genetics, 2012, 44, 319-322.	9.4	208
78	Unstable minisatellite expansion causing recessively inherited myoclonus epilepsy, EPM1. Nature Genetics, 1997, 15, 393-396.	9.4	207
79	Role of cancer-associated stromal fibroblasts in metastatic colon cancer to the liver and their expression profiles. Oncogene, 2004, 23, 7366-7377.	2.6	204
80	DNA mismatch repair gene mutations in 55 kindreds with verified or putative hereditary non-polyposis colorectal cancer. Human Molecular Genetics, 1996, 5, 763-769.	1.4	198
81	Mutations in a Novel Gene with Transmembrane Domains Underlie Usher Syndrome Type 3. American Journal of Human Genetics, 2001, 69, 673-684.	2.6	195
82	Molecular Analysis of Hereditary Nonpolyposis Colorectal Cancer in the United States: High Mutation Detection Rate among Clinically Selected Families and Characterization of an American Founder Genomic Deletion of the MSH2 Gene. American Journal of Human Genetics, 2003, 72, 1088-1100.	2.6	195
83	The 11q;22q translocation: A European collaborative analysis of 43 cases. Human Genetics, 1980, 56, 21-51.	1.8	192
84	Tumor formation and inactivation of RIZ1, an Rb-binding member of a nuclear protein-methyltransferase superfamily. Genes and Development, 2001, 15, 2250-2262.	2.7	181
85	Age-related hypermethylation of the 5' region of MLH1 in normal colonic mucosa is associated with microsatellite-unstable colorectal cancer development. Cancer Research, 2001, 61, 6991-5.	0.4	177
86	High BAALC expression associates with other molecular prognostic markers, poor outcome, and a distinct gene-expression signature in cytogenetically normal patients younger than 60 years with acute myeloid leukemia: a Cancer and Leukemia Group B (CALGB) study. Blood, 2008, 111, 5371-5379.	0.6	174
87	Rearrangement of ALL1 (MLL) in acute myeloid leukemia with normal cytogenetics. Cancer Research, 1998, 58, 55-9.	0.4	174
88	PTEN Mutational Spectra, Expression Levels, and Subcellular Localization in Microsatellite Stable and Unstable Colorectal Cancers. American Journal of Pathology, 2002, 161, 439-447.	1.9	173
89	Functional Significance and Clinical Phenotype of Nontruncating Mismatch Repair Variants of MLH1. Gastroenterology, 2005, 129, 537-549.	0.6	170
90	Effect of dystrophin gene deletions on mRNA levels and processing in Duchenne and Becker muscular dystrophies. Cell, 1990, 63, 1239-1248.	13.5	165

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91	Analytic review: nature and origin of males with XX sex chromosomes. American Journal of Human Genetics, 1972, 24, 71-105.	2.6	165
92	Aberrant expression of an amplified c-myb oncogene in two cell lines from a colon carcinoma.. Proceedings of the National Academy of Sciences of the United States of America, 1984, 81, 4534-4538.	3.3	163
93	Gelsolinâ€“derived familial amyloidosis caused by asparagine or tyrosine substitution for aspartic acid at residue 187. Nature Genetics, 1992, 2, 157-160.	9.4	163
94	Microsatellite Instability. New England Journal of Medicine, 2003, 349, 209-210.	13.9	160
95	Semiautomated assessment of loss of heterozygosity and replication error in tumors. Cancer Research, 1996, 56, 3331-7.	0.4	160
96	Gene encoding a new RING-B-box-Coiled-coil protein is mutated in mulibrey nanism. Nature Genetics, 2000, 25, 298-301.	9.4	159
97	The clinical significance of karyotype in acute myelogenous leukemia. Cancer Genetics and Cytogenetics, 1989, 40, 203-216.	1.0	157
98	Germline Allele-Specific Expression of TGFBR1 Confers an Increased Risk of Colorectal Cancer. Science, 2008, 321, 1361-1365.	6.0	157
99	Acute myeloid leukemia with complex karyotypes and abnormal chromosome 21: Amplification discloses overexpression of APP, ETS2, and ERG genes. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 3915-3920.	3.3	155
100	The Incidence of Lynch Syndrome. Familial Cancer, 2005, 4, 233-237.	0.9	154
101	The Frequency of Muir-Torre Syndrome Among Lynch Syndrome Families. Journal of the National Cancer Institute, 2008, 100, 277-281.	3.0	152
102	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	1.5	150
103	Mismatch Repair Gene PMS2. Cancer Research, 2004, 64, 4721-4727.	0.4	149
104	Mutations Predisposing to Hereditary Nonpolyposis Colorectal Cancer. Advances in Cancer Research, 1997, 71, 93-119.	1.9	148
105	Mutations in KERA, encoding keratocan, cause cornea plana. Nature Genetics, 2000, 25, 91-95.	9.4	148
106	Candidate tumor suppressor RIZ is frequently involved in colorectal carcinogenesis. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 2662-2667.	3.3	148
107	Cancer Risks for PMS2-Associated Lynch Syndrome. Journal of Clinical Oncology, 2018, 36, 2961-2968.	0.8	147
108	Phenotypic and genotypic heterogeneity in the Lynch syndrome: diagnostic, surveillance and management implications. European Journal of Human Genetics, 2006, 14, 390-402.	1.4	144

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109	Clinical Significance of Chromosomal Abnormalities in Acute Nonlymphoblastic Leukemia. Cancer Genetics and Cytogenetics, 1984, 11, 332-350.	1.0	140
110	Localization of the EPM1 gene for progressive myoclonus epilepsy on chromosome 21: linkage disequilibrium allows high resolution mapping. Human Molecular Genetics, 1993, 2, 1229-1234.	1.4	139
111	The Search for Unaffected Individuals with Lynch Syndrome: Do the Ends Justify the Means?. Cancer Prevention Research, 2011, 4, 1-5.	0.7	138
112	Linkage, physical mapping, and DNA sequence analysis of pseudoautosomal loci on the human X and Y chromosomes. Genomics, 1987, 1, 243-256.	1.3	137
113	Assessment of Tumor Sequencing as a Replacement for Lynch Syndrome Screening and Current Molecular Tests for Patients With Colorectal Cancer. JAMA Oncology, 2018, 4, 806.	3.4	136
114	Six-year follow-up of the clinical significance of karyotype in acute lymphoblastic leukemia. Cancer Genetics and Cytogenetics, 1989, 40, 171-185.	1.0	135
115	Y;autosome translocations and mosaicism in the aetiology of 45,X maleness: assignment of fertility factor to distal Yq11. Human Genetics, 1988, 79, 2-7.	1.8	134
116	BAALC, the human member of a novel mammalian neuroectoderm gene lineage, is implicated in hematopoiesis and acute leukemia. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 13901-13906.	3.3	133
117	A Susceptibility Locus for Papillary Thyroid Carcinoma on Chromosome 8q24. Cancer Research, 2009, 69, 625-631.	0.4	133
118	Amnionless, essential for mouse gastrulation, is mutated in recessive hereditary megaloblastic anemia. Nature Genetics, 2003, 33, 426-429.	9.4	132
119	Reconstructing hominid Y evolution: X-homologous block, created by X-Y transposition, was disrupted by Yp inversion through LINE--LINE recombination. Human Molecular Genetics, 1998, 7, 1-11.	1.4	131
120	Polymerase Î´ variants in RER colorectal tumours. Nature Genetics, 1995, 9, 10-11.	9.4	129
121	Assignment of the Muscle-Eye-Brain Disease Gene to 1p32-p34 by Linkage Analysis and Homozygosity Mapping. American Journal of Human Genetics, 1999, 64, 126-135.	2.6	128
122	In-Depth Characterization of the MicroRNA Transcriptome in Normal Thyroid and Papillary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1401-E1409.	1.8	125
123	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	2.6	124
124	Chromosome Y-specific DNA is transferred to the short arm of X chromosome in human XX males. Science, 1986, 233, 786-788.	6.0	123
125	Human type I procollagen genes are located on different chromosomes.. Proceedings of the National Academy of Sciences of the United States of America, 1982, 79, 6627-6630.	3.3	119
126	From the Cover: Loss of imprinting of the insulin-like growth factor II gene occurs by biallelic methylation in a core region of H19-associated CTCF-binding sites in colorectal cancer. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 591-596.	3.3	119

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127	Finnish hereditary amyloidosis is caused by a single nucleotide substitution in the gelsolin gene. FEBS Letters, 1990, 276, 75-77.	1.3	118
128	The Founder Mutation MSH2*1906Gâ†'C Is an Important Cause of Hereditary Nonpolyposis Colorectal Cancer in the Ashkenazi Jewish Population. American Journal of Human Genetics, 2002, 71, 1395-1412.	2.6	118
129	A genome-wide association study yields five novel thyroid cancer risk loci. Nature Communications, 2017, 8, 14517.	5.8	117
130	Predictive genetic testing for hereditary non-polyposis colorectal cancer: Uptake and long-term satisfaction. International Journal of Cancer, 2000, 89, 44-50.	2.3	116
131	MicroRNAs in Thyroid Cancer. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 3326-3336.	1.8	115
132	An abnormal terminal X-Y interchange accounts for most but not all cases of human XX maleness. Cell, 1987, 49, 595-602.	13.5	114
133	Cumulative Burden of Colorectal Cancerâ€"Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. Gastroenterology, 2020, 158, 1274-1286.e12.	0.6	110
134	Exchange of terminal portions of X- and Y-chromosomal short arms in human XX males. Nature, 1987, 328, 437-440.	13.7	109
135	Clonal Chromosomal Abnormalities Showing Multiple-Cell-Lineage Involvement in Acute Myeloid Leukemia. New England Journal of Medicine, 1988, 318, 1153-1158.	13.9	109
136	Monoamine oxidase deficiency in males with an X chromosome deletion. Neuron, 1989, 2, 1069-1076.	3.8	109
137	Expression of the human mismatch repair gene hMSH2 in normal and neoplastic tissues. Cancer Research, 1996, 56, 235-40.	0.4	109
138	MITOGENIC ACTION OF ANTILEUCOCYTE IMMUNE SERUM ON PERIPHERAL LEUCOCYTES IN VITRO. Lancet, The, 1963, 282, 385-386.	6.3	107
139	Erythroid cell development in fetal mice: Synthetic capacity for different proteins. Journal of Molecular Biology, 1968, 33, 79-91.	2.0	106
140	Epigenetic phenotypes distinguish microsatellite-stable and -unstable colorectal cancers. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 12661-12666.	3.3	105
141	Clinical-cytogenetic correlations in myelodysplasia (preleukemia). Cancer Genetics and Cytogenetics, 1989, 40, 149-161.	1.0	104
142	Pericentric inversions of human chromosomes 9 and 10. American Journal of Human Genetics, 1974, 26, 746-66.	2.6	104
143	Downregulated in adenoma gene encodes a chloride transporter defective in congenital chloride diarrhea. American Journal of Physiology - Renal Physiology, 1999, 276, G185-G192.	1.6	103
144	Genetic Testing for Cancer Predisposition. Annual Review of Medicine, 2001, 52, 371-400.	5.0	103

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145	Trisomy 12 in B Cells of Patients with B-Cell Chronic Lymphocytic Leukemia. New England Journal of Medicine, 1986, 314, 865-869.	13.9	101
146	Molecular mapping of the putative gonadoblastoma locus on the Y chromosome. Genes Chromosomes and Cancer, 1995, 14, 210-214.	1.5	101
147	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. Nature, 2020, 586, 769-775.	13.7	101
148	Chromosome Y-specific DNA in related human XX males. Nature, 1985, 315, 224-226.	13.7	100
149	Monosomy 7 in Granulocytes and Monocytes in Myelodysplastic Syndrome. New England Journal of Medicine, 1987, 316, 499-503.	13.9	100
150	MSH2 andMLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. , 1997, 18, 269-278.		99
151	Hereditary juvenile cobalamin deficiency caused by mutations in the intrinsic factor gene. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 4130-4133.	3.3	97
152	Comprehensive population-wide analysis of Lynch syndrome in Iceland reveals founder mutations in MSH6 and PMS2. Nature Communications, 2017, 8, 14755.	5.8	96
153	Genetic Predisposition to Papillary Thyroid Carcinoma: Involvement of FOXE1, TSHR, and a Novel lincRNA Gene, PTCSC2. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E164-E172.	1.8	93
154	MicroRNA Signature in Thyroid Fine Needle Aspiration Cytology Applied to â€œAtypia of Undetermined Significanceâ€•Cases. Thyroid, 2012, 22, 9-16.	2.4	92
155	Hypermethylation, but not LOH, is associated with the low expression ofMT1G andCRABP1 in papillary thyroid carcinoma. International Journal of Cancer, 2003, 104, 735-744.	2.3	91
156	Long-range PCR facilitates the identification ofPMS2-specific mutations. Human Mutation, 2006, 27, 490-495.	1.1	90
157	Circulating Levels of Insulin-like Growth Factor 1 and Insulin-like Growth Factor Binding Protein 3 Associate With Risk of Colorectal Cancer Based on Serologic and Mendelian Randomization Analyses. Gastroenterology, 2020, 158, 1300-1312.e20.	0.6	90
158	Genetic evidence of Xâ€“Y interchange in a human XX male. Nature, 1984, 307, 170-171.	13.7	89
159	Peutz-Jeghers disease: most, but not all, families are compatible with linkage to 19p13.3.. Journal of Medical Genetics, 1998, 35, 42-44.	1.5	89
160	Polymorphic Variation at the BAT-25 and BAT-26 Loci in Individuals of African Origin. American Journal of Pathology, 1999, 155, 349-353.	1.9	89
161	Microsatellite Instability in Adenomas as a Marker for Hereditary Nonpolyposis Colorectal Cancer. American Journal of Pathology, 1999, 155, 1849-1853.	1.9	89
162	Functional Significance and Clinical Phenotype of Nontruncating Mismatch Repair Variants of. Gastroenterology, 2005, 129, 537-549.	0.6	89

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163	Comment on: Screening for Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer) among Endometrial Cancer Patients. Cancer Research, 2007, 67, 9603-9603.	0.4	88
164	Prostate cancer incidence in males with Lynch syndrome. Genetics in Medicine, 2014, 16, 553-557.	1.1	88
165	Somatic Acquisition and Signaling of <EMPH TYPE="ITAL">TGFBR1</EMPH>*6A in Cancer. JAMA - Journal of the American Medical Association, 2005, 294, 1634.	3.8	87
166	Biallelic MUTYH mutations can mimic Lynch syndrome. European Journal of Human Genetics, 2014, 22, 1334-1337.	1.4	87
167	Recessive sex-determining genes in human XX male syndrome. Cell, 1978, 15, 837-842.	13.5	86
168	Abnormalities of chromosome No. 17 in myeloproliferative disorders. Cancer Genetics and Cytogenetics, 1982, 5, 123-135.	1.0	86
169	Familial erythrocytosis genetically linked to erythropoietin receptor gene. Lancet, The, 1993, 341, 82-84.	6.3	86
170	GWAS of thyroid stimulating hormone highlights pleiotropic effects and inverse association with thyroid cancer. Nature Communications, 2020, 11, 3981.	5.8	86
171	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, 6054-6058.	3.3	85
172	Nonlinear partial differential equations and applications: Gene expression profiling of isogenic cells with different TP53 gene dosage reveals numerous genes that are affected by TP53 dosage and identifies CSPG2 as a direct target of p53. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15632-15637.	3.3	85
173	Cartilage-hair hypoplasia gene assigned to chromosome 9 by linkage analysis. Nature Genetics, 1993, 3, 338-341.	9.4	84
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