Albert de la Chapelle

List of Publications by Year
in descending order

472	79,628	799 115	505 262
papers	citations	h-index	g-index

595	595	595	44328
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.	29.6	7,962
2	Revised Bethesda Guidelines for Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome) and Microsatellite Instability. Journal of the National Cancer Institute, 2004, 96, 261-268.	6.2	2,796
3	Mutations of a mutS homolog in hereditary nonpolyposis colorectal cancer. Cell, 1993, 75, 1215-1225.	27.3	2,200
4	Hereditary Colorectal Cancer. New England Journal of Medicine, 2003, 348, 919-932.	29.6	1,887
5	Mutations of two P/WS homologues in hereditary nonpolyposis colon cancer. Nature, 1994, 371, 75-80.	35.3	1,528
6	A serine/threonine kinase gene defective in Peutzâ€“Jeghers syndrome. Nature, 1998, 391, 184-187.	35.3	1,466
7	Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. Gastroenterology, 2000, 118, 829-834.	1.3	1,265
8	Screening for the Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer). New England Journal of Medicine, 2005, 352, 1851-1860.	29.6	1,258
9	Diverse spermatogenic defects in humans caused by Y chromosome deletions encompassing a novel RNAâ€“binding protein gene. Nature Genetics, 1995, 10, 383-393.	20.1	1,187
10	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.	7.4	1,144
11	Cancer risk in mutation carriers of DNA-mismatch-repair genes. International Journal of Cancer, 1999, 81, 214-218.	5.3	1,070
12	Incidence of Hereditary Nonpolyposis Colorectal Cancer and the Feasibility of Molecular Screening for the Disease. New England Journal of Medicine, 1998, 338, 1481-1487.	29.6	1,055
13	Hypermutability and mismatch repair deficiency in RER+ tumor cells. Cell, 1993, 75, 1227-1236.	27.3	1,034
14	Mutation in the follicle-stimulating hormone receptor gene causes hereditary hypergonadotropic ovarian failure. Cell, 1995, 82, 959-968.	27.3	909
15	Analysis of mismatch repair genes in hereditary nonâ€“polyposis colorectal cancer patients. Nature Medicine, 1996, 2, 169-174.	29.5	896
16	The sex-determining region of the human Y chromosome encodes a finger protein. Cell, 1987, 51, 1091-1104.	27.3	882
17	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.	7.4	801
18	Feasibility of Screening for Lynch Syndrome Among Patients With Colorectal Cancer. Journal of Clinical Oncology, 2008, 26, 5783-5788.	5.7	774

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19	Mismatch repair gene defects in sporadic colorectal cancers with microsatellite instability. Nature Genetics, 1995, 9, 48-55.	20.1	760
20	The diastrophic dysplasia gene encodes a novel sulfate transporter: Positional cloning by fine-structure linkage disequilibrium mapping. Cell, 1994, 78, 1073-1087.	27.3	734
21	Xâ€“linked anhidrotic (hypohidrotic) ectodermal dysplasia is caused by mutation in a novel transmembrane protein. Nature Genetics, 1996, 13, 409-416.	20.1	698
22	Genetic predisposition to colorectal cancer. Nature Reviews Cancer, 2004, 4, 769-780.	28.2	569
23	Linkage disequilibrium mapping in isolated founder populations: diastrophic dysplasia in Finland. Nature Genetics, 1992, 2, 204-211.	20.1	545
24	Prevalence and Spectrum of Germline Cancer Susceptibility Gene Mutations Among Patients With Early-Onset Colorectal Cancer. JAMA Oncology, 2017, 3, 464.	7.2	533
25	The Clinical Phenotype of Lynch Syndrome Due to Germ-Line PMS2 Mutations. Gastroenterology, 2008, 135, 419-428.e1.	1.3	489
26	Loss-of-Function Mutations in PPARÎ³ Associated with Human Colon Cancer. Molecular Cell, 1999, 3, 799-804.	9.4	487
27	Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. Nature, 1986, 322, 73-77.	35.3	476
28	Population-Based Molecular Detection of Hereditary Nonpolyposis Colorectal Cancer. Journal of Clinical Oncology, 2000, 18, 2193-2200.	5.7	468
29	Mutations in the RNA Component of RNase MRP Cause a Pleiotropic Human Disease, Cartilage-Hair Hypoplasia. Cell, 2001, 104, 195-203.	27.3	467
30	Identification of Lynch Syndrome Among Patients With Colorectal Cancer. JAMA - Journal of the American Medical Association, 2012, 308, 1555.	6.9	453
31	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.	20.1	448
32	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	20.1	409
33	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.	7.4	399
34	Mutations of the Downâ€“regulated in adenoma (DRA) gene cause congenital chloride diarrhoea. Nature Genetics, 1996, 14, 316-319.	20.1	396
35	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.	7.4	375
36	A deletion in chromosome 22 can cause digeorge syndrome. Human Genetics, 1981, 57, 253-256.	3.8	364

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37	Common variants on 9q22.33 and 14q13.3 predispose to thyroid cancer in European populations. Nature Genetics, 2009, 41, 460-464.	20.1	355
38	Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer Syndrome: Later Age of Onset. Gastroenterology, 2005, 129, 415-421.	1.3	340
39	Downregulation of Death-Associated Protein Kinase 1 (DAPK1) in Chronic Lymphocytic Leukemia. Cell, 2007, 129, 879-890.	27.3	339
40	Colon and Endometrial Cancers With Mismatch Repair Deficiency Can Arise From Somatic, Rather Than Germline, Mutations. Gastroenterology, 2014, 147, 1308-1316.e1.	1.3	337
41	Risks of Lynch Syndrome Cancers for MSH6 Mutation Carriers. Journal of the National Cancer Institute, 2010, 102, 193-201.	6.2	334
42	Reprogramming of miRNA networks in cancer and leukemia. Genome Research, 2010, 20, 589-599.	5.5	331
43	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.	6.0	329
44	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.	7.4	313
45	Cancer Risk in Hereditary Nonpolyposis Colorectal Cancer Syndrome: Later Age of Onset. Gastroenterology, 2005, 129, 415-421.	1.3	312
46	Loss of the wild type MLH1 gene is a feature of hereditary nonpolyposis colorectal cancer. Nature Genetics, 1994, 8, 405-410.	20.1	309
47	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.	7.4	307
48	Disease gene mapping in isolated human populations: the example of Finland.. Journal of Medical Genetics, 1993, 30, 857-865.	3.5	303
49	Cubilin dysfunction causes abnormal metabolism of the steroid hormone 25(OH) vitamin D ‹sub›3‹/sub›. Proceedings of the National Academy of Sciences of the United States of America, 2001, 98, 13895-13900.	7.4	292
50	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.	7.4	291
51	Clinical Relevance of Microsatellite Instability in Colorectal Cancer. Journal of Clinical Oncology, 2010, 28, 3380-3387.	5.7	281
52	The neuronal ceroid lipofuscinoses in human EPMR and mnd mutant mice are associated with mutations in CLN8. Nature Genetics, 1999, 23, 233-236.	20.1	279
53	Founding mutations and Alu-mediated recombination in hereditary colon cancer. Nature Medicine, 1995, 1, 1203-1206.	29.5	276
54	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.	7.4	267

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55	The functional cobalamin (vitamin B12)â€“intrinsic factor receptor is a novel complex of cubilin and amnionless. Blood, 2004, 103, 1573-1579.	1.4	263
56	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.	5.6	263
57	Genetic and Epigenetic Modification of MLH1 Accounts for a Major Share of Microsatellite-Unstable Colorectal Cancers. American Journal of Pathology, 2000, 156, 1773-1779.	4.0	257
58	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.	29.5	251
59	Conversion of diploidy to haploidy. Nature, 2000, 403, 723-724.	35.3	248
60	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.	7.4	241
61	Mutations in CUBN, encoding the intrinsic factor-vitamin B12 receptor, cubilin, cause hereditary megaloblastic anaemia 1. Nature Genetics, 1999, 21, 309-313.	20.1	237
62	Assignment of an Usher syndrome type III (USH3) gene to chromosome 3q. Human Molecular Genetics, 1995, 4, 93-98.	3.0	228
63	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.	5.7	226
64	The etiology of maleness in XX men. Human Genetics, 1981, 58, 105-116.	3.8	225
65	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.	1.4	222
66	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.	7.4	214
67	Pseudoautosomal DNA sequences in the pairing region of the human sex chromosomes. Nature, 1985, 317, 692-697.	35.3	212
68	Discovery of common variants associated with low TSH levels and thyroid cancer risk. Nature Genetics, 2012, 44, 319-322.	20.1	211
69	Unstable minisatellite expansion causing recessively inherited myoclonus epilepsy, EPM1. Nature Genetics, 1997, 15, 393-396.	20.1	207
70	Role of cancer-associated stromal fibroblasts in metastatic colon cancer to the liver and their expression profiles. Oncogene, 2004, 23, 7366-7377.	5.8	205
71	DNA mismatch repair gene mutations in 55 kindreds with verified or putative hereditary non-polyposis colorectal cancer. Human Molecular Genetics, 1996, 5, 763-769.	3.0	198
72	Mutations in a Novel Gene with Transmembrane Domains Underlie Usher Syndrome Type 3. American Journal of Human Genetics, 2001, 69, 673-684.	6.0	197

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73	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.	6.0	195
74	The 11q;22q translocation: A European collaborative analysis of 43 cases. Human Genetics, 1980, 56, 21-51.	3.8	193
75	Tumor formation and inactivation of RIZ1, an Rb-binding member of a nuclear protein-methyltransferase superfamily. Genes and Development, 2001, 15, 2250-2262.	5.8	181
76	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.	1.4	175
77	PTEN Mutational Spectra, Expression Levels, and Subcellular Localization in Microsatellite Stable and Unstable Colorectal Cancers. American Journal of Pathology, 2002, 161, 439-447.	4.0	174
78	Functional Significance and Clinical Phenotype of Nontruncating Mismatch Repair Variants of MLH1. Gastroenterology, 2005, 129, 537-549.	1.3	170
79	Effect of dystrophin gene deletions on mRNA levels and processing in Duchenne and Becker muscular dystrophies. Cell, 1990, 63, 1239-1248.	27.3	165
80	Gelsolinâ€“derived familial amyloidosis caused by asparagine or tyrosine substitution for aspartic acid at residue 187. Nature Genetics, 1992, 2, 157-160.	20.1	165
81	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.	7.4	163
82	Gene encoding a new RING-B-box-Coiled-coil protein is mutated in mulibrey nanism. Nature Genetics, 2000, 25, 298-301.	20.1	161
83	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	3.3	161
84	Germline Allele-Specific Expression of TGFBR1 Confers an Increased Risk of Colorectal Cancer. Science, 2008, 321, 1361-1365.	19.6	158
85	The clinical significance of karyotype in acute myelogenous leukemia. Cancer Genetics and Cytogenetics, 1989, 40, 203-216.	0.9	157
86	The Incidence of Lynch Syndrome. Familial Cancer, 2005, 4, 233-237.	2.0	157
87	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.	7.4	156
88	The Frequency of Muir-Torre Syndrome Among Lynch Syndrome Families. Journal of the National Cancer Institute, 2008, 100, 277-281.	6.2	155
89	Mutations in KERA, encoding keratocan, cause cornea plana. Nature Genetics, 2000, 25, 91-95.	20.1	150
90	Mutations Predisposing to Hereditary Nonpolyposis Colorectal Cancer. Advances in Cancer Research, 1997, 71, 93-119.	6.3	149

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91	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.	7.4	148
92	Phenotypic and genotypic heterogeneity in the Lynch syndrome: diagnostic, surveillance and management implications. European Journal of Human Genetics, 2006, 14, 390-402.	2.8	144
93	The Search for Unaffected Individuals with Lynch Syndrome: Do the Ends Justify the Means?. Cancer Prevention Research, 2011, 4, 1-5.	1.5	142
94	Clinical Significance of Chromosomal Abnormalities in Acute Nonlymphoblastic Leukemia. Cancer Genetics and Cytogenetics, 1984, 11, 332-350.	0.9	140
95	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.	7.2	140
96	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.	3.0	139
97	Linkage, physical mapping, and DNA sequence analysis of pseudoautosomal loci on the human X and Y chromosomes. Genomics, 1987, 1, 243-256.	2.9	137
98	Six-year follow-up of the clinical significance of karyotype in acute lymphoblastic leukemia. Cancer Genetics and Cytogenetics, 1989, 40, 171-185.	0.9	135
99	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	6.0	135
100	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.	3.8	134
101	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.	7.4	133
102	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.	3.0	132
103	Amnionless, essential for mouse gastrulation, is mutated in recessive hereditary megaloblastic anemia. Nature Genetics, 2003, 33, 426-429.	20.1	132
104	In-Depth Characterization of the MicroRNA Transcriptome in Normal Thyroid and Papillary Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1401-E1409.	3.5	130
105	Polymerase Î´ variants in RER colorectal tumours. Nature Genetics, 1995, 9, 10-11.	20.1	129
106	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.	6.0	128
107	A genome-wide association study yields five novel thyroid cancer risk loci. Nature Communications, 2017, 8, 14517.	12.8	127
108	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.	7.4	119

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109	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.	1.3	119
110	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.	7.4	119
111	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.	6.0	118
112	Predictive genetic testing for hereditary non-polyposis colorectal cancer: Uptake and long-term satisfaction. International Journal of Cancer, 2000, 89, 44-50.	5.3	117
113	MicroRNAs in Thyroid Cancer. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 3326-3336.	3.5	117
114	An abnormal terminal X-Y interchange accounts for most but not all cases of human XX maleness. Cell, 1987, 49, 595-602.	27.3	114
115	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. Nature, 2020, 586, 769-775.	35.3	110
116	Exchange of terminal portions of X- and Y-chromosomal short arms in human XX males. Nature, 1987, 328, 437-440.	35.3	109
117	Clonal Chromosomal Abnormalities Showing Multiple-Cell-Lineage Involvement in Acute Myeloid Leukemia. New England Journal of Medicine, 1988, 318, 1153-1158.	29.6	109
118	Monoamine oxidase deficiency in males with an X chromosome deletion. Neuron, 1989, 2, 1069-1076.	7.9	109
119	MITOGENIC ACTION OF ANTILEUCOCYTE IMMUNE SERUM ON PERIPHERAL LEUCOCYTES IN VITRO. Lancet, The, 1963, 282, 385-386.	11.9	107
120	Erythroid cell development in fetal mice: Synthetic capacity for different proteins. Journal of Molecular Biology, 1968, 33, 79-91.	4.2	106
121	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.	7.4	105
122	Clinical-cytogenetic correlations in myelodysplasia (preleukemia). Cancer Genetics and Cytogenetics, 1989, 40, 149-161.	0.9	104
123	Downregulated in adenoma gene encodes a chloride transporter defective in congenital chloride diarrhea. American Journal of Physiology - Renal Physiology, 1999, 276, G185-G192.	3.5	104
124	Genetic Testing for Cancer Predisposition. Annual Review of Medicine, 2001, 52, 371-400.	11.9	103
125	Trisomy 12 in B Cells of Patients with B-Cell Chronic Lymphocytic Leukemia. New England Journal of Medicine, 1986, 314, 865-869.	29.6	101
126	Molecular mapping of the putative gonadoblastoma locus on the Y chromosome. Genes Chromosomes and Cancer, 1995, 14, 210-214.	3.2	101

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127	Chromosome Y-specific DNA in related human XX males. Nature, 1985, 315, 224-226.	35.3	100
128	Monosomy 7 in Granulocytes and Monocytes in Myelodysplastic Syndrome. New England Journal of Medicine, 1987, 316, 499-503.	29.6	100
129	MSH2 andMLH1 mutations in sporadic replication error-positive colorectal carcinoma as assessed by two-dimensional DNA electrophoresis. Genes Chromosomes and Cancer, 1997, 18, 269-278.	3.2	100
130	Comprehensive population-wide analysis of Lynch syndrome in Iceland reveals founder mutations in MSH6 and PMS2. Nature Communications, 2017, 8, 14755.	12.8	99
131	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.	7.4	98
132	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.	1.3	98
133	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.	3.5	95
134	GWAS of thyroid stimulating hormone highlights pleiotropic effects and inverse association with thyroid cancer. Nature Communications, 2020, 11, 3981.	12.8	95
135	Long-range PCR facilitates the identification ofPMS2-specific mutations. Human Mutation, 2006, 27, 490-495.	2.7	94
136	Hypermethylation, but not LOH, is associated with the low expression ofMT1G andCRABP1 in papillary thyroid carcinoma. International Journal of Cancer, 2003, 104, 735-744.	5.3	93
137	Biallelic MUTYH mutations can mimic Lynch syndrome. European Journal of Human Genetics, 2014, 22, 1334-1337.	2.8	93
138	MicroRNA Signature in Thyroid Fine Needle Aspiration Cytology Applied to â€œAtypia of Undetermined Significanceâ€•Cases. Thyroid, 2012, 22, 9-16.	4.8	92
139	Adiposity, metabolites, and colorectal cancer risk: Mendelian randomization study. BMC Medicine, 2020, 18, 396.	5.6	92
140	Prostate cancer incidence in males with Lynch syndrome. Genetics in Medicine, 2014, 16, 553-557.	2.4	91
141	Genetic evidence of Xâ€“Y interchange in a human XX male. Nature, 1984, 307, 170-171.	35.3	89
142	Peutz-Jeghers disease: most, but not all, families are compatible with linkage to 19p13.3.. Journal of Medical Genetics, 1998, 35, 42-44.	3.5	89
143	Polymorphic Variation at the BAT-25 and BAT-26 Loci in Individuals of African Origin. American Journal of Pathology, 1999, 155, 349-353.	4.0	89
144	Microsatellite Instability in Adenomas as a Marker for Hereditary Nonpolyposis Colorectal Cancer. American Journal of Pathology, 1999, 155, 1849-1853.	4.0	89

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145	Functional Significance and Clinical Phenotype of Nontruncating Mismatch Repair Variants of. Gastroenterology, 2005, 129, 537-549.	1.3	89
146	Familial erythrocytosis genetically linked to erythropoietin receptor gene. Lancet, The, 1993, 341, 82-84.	11.9	88
147	Somatic Acquisition and Signaling of <EMPH TYPE="ITAL">TGFBR1</EMPH>*6A in Cancer. JAMA - Journal of the American Medical Association, 2005, 294, 1634.	6.9	88
148	Comment on: Screening for Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer) among Endometrial Cancer Patients. Cancer Research, 2007, 67, 9603-9603.	0.9	88
149	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.	7.4	87
150	Recessive sex-determining genes in human XX male syndrome. Cell, 1978, 15, 837-842.	27.3	86
151	Abnormalities of chromosome No. 17 in myeloproliferative disorders. Cancer Genetics and Cytogenetics, 1982, 5, 123-135.	0.9	86
152	Clinical and genetic heterogeneity in autosomal recessive nemaline myopathy. Neuromuscular Disorders, 1999, 9, 564-572.	0.7	86
153	Aberrant splicing of the CHM gene is a significant cause of choroideremia. Nature Genetics, 1992, 1, 109-113.	20.1	85
154	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.	7.4	85
155	A germline mutation in SRRM2, a splicing factor gene, is implicated in papillary thyroid carcinoma predisposition. Scientific Reports, 2015, 5, 10566.	3.4	85
156	Cartilage-hair hypoplasia gene assigned to chromosome 9 by linkage analysis. Nature Genetics, 1993, 3, 338-341.	20.1	84
157	Genetic Control of Serum IgE Levels and Asthma: Linkage and Linkage Disequilibrium Studies in an Isolated Population. Human Molecular Genetics, 1997, 6, 2069-2076.	3.0	83
158	Thyroid Hormone Receptor Î² (THRB) Is a Major Target Gene for MicroRNAs Deregulated in Papillary Thyroid Carcinoma (PTC). Journal of Clinical Endocrinology and Metabolism, 2011, 96, E546-E553.	3.5	83
159	Mutation patterns identify adult patients with de novo acute myeloid leukemia aged 60 years or older who respond favorably to standard chemotherapy: an analysis of Alliance studies. Leukemia, 2018, 32, 1338-1348.	7.3	83
160	Defective chemotaxis in monosomy-7. Nature, 1977, 265, 146-147.	35.3	82
161	Cohen syndrome gene assigned to the long arm of chromosome 8 by linkage analysis. Nature Genetics, 1994, 7, 201-204.	20.1	81
162	Cancer Risks for Relatives of Patients With Serrated Polyposis. American Journal of Gastroenterology, 2012, 107, 770-778.	0.4	81

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163	The distribution of linkage disequilibrium over anonymous genome regions. Human Molecular Genetics, 1995, 4, 887-894.	3.0	80
164	Genetic Background of Congenital Chloride Diarrhea in High-Incidence Populations: Finland, Poland, and Saudi Arabia and Kuwait. American Journal of Human Genetics, 1998, 63, 760-768.	6.0	80
165	MYH9 binds to lncRNA gene PTCSC2 and regulates FOXE1 in the 9q22 thyroid cancer risk locus. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 474-479.	7.4	80
166	Multiple functional variants in long-range enhancer elements contribute to the risk of SNP rs965513 in thyroid cancer. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 6128-6133.	7.4	79
167	Genetics of Hereditary Colon Cancer. Annual Review of Genetics, 1995, 29, 329-348.	7.7	78
168	HÃ†MOPHILIA A IN A GIRL. Lancet, The, 1961, 278, 578-580.	11.9	76
169	Clozapine-induced agranulocytosis. Human Genetics, 1977, 37, 183-194.	3.8	76
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