Charis Eng

List of Publications by Year
in descending order

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621 papers	59,902 citations	701 121 h-index	1424 221 g-index
637 all docs	637 docs citations	637 times ranked	40129 citing authors

#	Article	IF	Citations
1	Germline mutations of the PTEN gene in Cowden disease, an inherited breast and thyroid cancer syndrome. Nature Genetics, 1997, 16, 64-67.	21.4	1,902
2	Germ-line mutations of the RET proto-oncogene in multiple endocrine neoplasia type 2A. Nature, 1993, 363, 458-460.	27.8	1,886
3	Germ-Line Mutations in Nonsyndromic Pheochromocytoma. New England Journal of Medicine, 2002, 346, 1459-1466.	27.0	1,299
4	Medullary Thyroid Cancer: Management Guidelines of the American Thyroid Association. Thyroid, 2009, 19, 565-612.	4.5	1,247
5	Gene Mutations in the Succinate Dehydrogenase Subunit SDHB Cause Susceptibility to Familial Pheochromocytoma and to Familial Paraganglioma. American Journal of Human Genetics, 2001, 69, 49-54.	6.2	1,021
6	Catalytic specificity of protein-tyrosine kinases is critical for selective signalling. Nature, 1995, 373, 536-539.	27.8	932
7	Essential Role for Nuclear PTEN in Maintaining Chromosomal Integrity. Cell, 2007, 128, 157-170.	28.9	879
8	Distinct Clinical Features of Paraganglioma Syndromes Associated With <EMPH TYPE="ITAL">SDHB</EMPH> and <EMPH TYPE="ITAL">SDHD</EMPH> Gene Mutations. JAMA - Journal of the American Medical Association, 2004, 292, 943.	7.4	821
9	P-TEN, the tumor suppressor from human chromosome 10q23, is a dual-specificity phosphatase. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 9052-9057.	7.1	765
10	Lifetime Cancer Risks in Individuals with Germline PTEN Mutations. Clinical Cancer Research, 2012, 18, 400-407.	7.0	738
11	PTEN: One Gene, Many Syndromes. Human Mutation, 2003, 22, 183-198.	2.5	729
12	Altered PTEN Expression as a Diagnostic Marker for the Earliest Endometrial Precancers. Journal of the National Cancer Institute, 2000, 92, 924-930.	6.3	709
13	Subset of individuals with autism spectrum disorders and extreme macrocephaly associated with germline PTEN tumour suppressor gene mutations. Journal of Medical Genetics, 2005, 42, 318-321.	3.2	673
14	Specific mutations of the RET proto-oncogene are related to disease phenotype in MEN 2A and FMTC. Nature Genetics, 1994, 6, 70-74.	21.4	647
15	Localization of the gene for Cowden disease to chromosome 10q22â€“23. Nature Genetics, 1996, 13, 114-116.	21.4	630
16	Germline mutations in PTEN are present in Bannayan-Zonana syndrome. Nature Genetics, 1997, 16, 333-334.	21.4	622
17	Mutation spectrum and genotype-phenotype analyses in Cowden disease and Bannayan-Zonana syndrome, two hamartoma syndromes with germline PTEN mutation. Human Molecular Genetics, 1998, 7, 507-515.	2.9	578
18	PTEN Mutation Spectrum and Genotype-Phenotype Correlations in Bannayan-Riley-Ruvalcaba Syndrome Suggest a Single Entity With Cowden Syndrome. Human Molecular Genetics, 1999, 8, 1461-1472.	2.9	562

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19	Mortality From Second Tumors Among Long-Term Survivors of Retinoblastoma. Journal of the National Cancer Institute, 1993, 85, 1121-1128.	6.3	553
20	Point mutation within the tyrosine kinase domain of the RET proto-oncogene in multiple endocrine neoplasia type 2B and related sporadic tumours. Human Molecular Genetics, 1994, 3, 237-241.	2.9	540
21	The relationship between specific RET proto-oncogene mutations and disease phenotype in multiple endocrine neoplasia type 2. International RET mutation consortium analysis. JAMA - Journal of the American Medical Association, 1996, 276, 1575-1579.	7.4	516
22	Loss-of-Function Mutations in PPARÎ³ Associated with Human Colon Cancer. Molecular Cell, 1999, 3, 799-804.	9.7	485
23	Will the real Cowden syndrome please stand up: revised diagnostic criteria. Journal of Medical Genetics, 2000, 37, 828-830.	3.2	483
24	Implementing genomic medicine in the clinic: the future is here. Genetics in Medicine, 2013, 15, 258-267.	2.4	472
25	Clinical and molecular genetics of patients with the Carneyâ€“Stratakis syndrome and germline mutations of the genes coding for the succinate dehydrogenase subunits SDHB, SDHC, and SDHD. European Journal of Human Genetics, 2008, 16, 79-88.	2.8	446
26	Pheochromocytoma and Paraganglioma. New England Journal of Medicine, 2019, 381, 552-565.	27.0	437
27	Protean PTEN: Form and Function. American Journal of Human Genetics, 2002, 70, 829-844.	6.2	432
28	Mutation of the endothelin-3 gene in the Waardenburg-Hirschsprung disease (Shah-Waardenburg) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	21.4	425
29	Methylation of the CDH1 promoter as the second genetic hit in hereditary diffuse gastric cancer. Nature Genetics, 2000, 26, 16-17.	21.4	420
30	Frequent somatic mutations in PTEN and TP53 are mutually exclusive in the stroma of breast carcinomas. Nature Genetics, 2002, 32, 355-357.	21.4	402
31	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.	7.1	399
32	Early-Onset Renal Cell Carcinoma as a Novel Extraparaganglial Component of SDHB-Associated Heritable Paraganglioma. American Journal of Human Genetics, 2004, 74, 153-159.	6.2	367
33	A role for mitochondrial enzymes in inherited neoplasia and beyond. Nature Reviews Cancer, 2003, 3, 193-202.	28.4	359
34	A Clinical Scoring System for Selection of Patients for PTEN Mutation Testing Is Proposed on the Basis of a Prospective Study of 3042 Probands. American Journal of Human Genetics, 2011, 88, 42-56.	6.2	332
35	Direct Evidence for Epithelial-Mesenchymal Transitions in Breast Cancer. Cancer Research, 2008, 68, 937-945.	0.9	329
36	TheRETProto-Oncogene in Multiple Endocrine Neoplasia Type 2 and Hirschsprung's Disease. New England Journal of Medicine, 1996, 335, 943-951.	27.0	317

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37	PTEN hamartoma tumor syndrome: An overview. Genetics in Medicine, 2009, 11, 687-694.	2.4	311
38	Predictors and Prevalence of Paraganglioma Syndrome Associated With Mutations of the <EMPH TYPE="ITAL">SDHC</EMPH> Gene. JAMA - Journal of the American Medical Association, 2005, 294, 2057.	7.4	309
39	New insights into genetic susceptibility of COVID-19: an ACE2 and TMPRSS2 polymorphism analysis. BMC Medicine, 2020, 18, 216.	5.5	304
40	Highly penetrant hereditary cancer syndromes. Oncogene, 2004, 23, 6445-6470.	5.9	302
41	PTEN mutations in gliomas and glioneuronal tumors. Oncogene, 1998, 16, 2259-2264.	5.9	300
42	The relationship between specific RET proto-oncogene mutations and disease phenotype in multiple endocrine neoplasia type 2. International RET mutation consortium analysis. JAMA - Journal of the American Medical Association, 1996, 276, 1575-9.	7.4	289
43	Breast-Cancer Stromal Cells withTP53Mutations and Nodal Metastases. New England Journal of Medicine, 2007, 357, 2543-2551.	27.0	288
44	Validation of Proposed DSM-5 Criteria for Autism Spectrum Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2012, 51, 28-40.e3.	0.5	287
45	A Limited Set of Human MicroRNA Is Deregulated in Follicular Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 3584-3591.	3.6	285
46	Differential Nuclear and Cytoplasmic Expression of PTEN in Normal Thyroid Tissue, and Benign and Malignant Epithelial Thyroid Tumors. American Journal of Pathology, 2000, 156, 1693-1700.	3.8	283
47	Germline PTEN Promoter Mutations and Deletions in Cowden/Bannayan-Riley-Ruvalcaba Syndrome Result in Aberrant PTEN Protein and Dysregulation of the Phosphoinositol-3-Kinase/Akt Pathway. American Journal of Human Genetics, 2003, 73, 404-411.	6.2	283
48	Will the real Cowden syndrome please stand up (again)? Expanding mutational and clinical spectra of the PTEN hamartoma tumour syndrome. Journal of Medical Genetics, 2004, 41, 323-326.	3.2	282
49	The nuclear affairs of PTEN. Journal of Cell Science, 2008, 121, 249-253.	2.0	259
50	From developmental disorder to heritable cancer: it's all in the BMP/TGF-Î² family. Nature Reviews Genetics, 2003, 4, 763-773.	16.3	258
51	PTEN is inversely correlated with the cell survival factor Akt/PKB and is inactivated via multiple mechanismsin haematological malignancies. Human Molecular Genetics, 1999, 8, 185-193.	2.9	254
52	Epigenetic PTEN Silencing in Malignant Melanomas without PTEN Mutation. American Journal of Pathology, 2000, 157, 1123-1128.	3.8	254
53	APC-dependent suppression of colon carcinogenesis by PPARÎ³. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 13771-13776.	7.1	252
54	Frequent Gastrointestinal Polyps and Colorectal Adenocarcinomas in a Prospective Series of PTEN Mutation Carriers. Gastroenterology, 2010, 139, 1927-1933.	1.3	251

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55	Diverse phenotypes associated with exon 10 mutations of the RET proto-oncogene. Human Molecular Genetics, 1994, 3, 2163-2168.	2.9	239
56	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, 704-711.	6.2	236
57	Germline SDHD mutation in familial phaeochromocytoma. Lancet, The, 2001, 357, 1181-1182.	13.7	236
58	PTEN coordinates G1 arrest by down-regulating cyclin D1 via its protein phosphatase activity and up-regulating p27 via its lipid phosphatase activity in a breast cancer model. Human Molecular Genetics, 2001, 10, 599-604.	2.9	235
59	Cancer phenomics: RET and PTEN as illustrative models. Nature Reviews Cancer, 2007, 7, 35-45.	28.4	231
60	Molecular Classification of Patients With Unexplained Hamartomatous and Hyperplastic Polyposis. JAMA - Journal of the American Medical Association, 2005, 294, 2465.	7.4	218
61	Germline Dinucleotide Mutation in Codon 883 of theRETProto-Oncogene in Multiple Endocrine Neoplasia Type 2B Without Codon 918 Mutation. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 3902-3904.	3.6	216
62	Hypomethylation of Noncoding DNA Regions and Overexpression of the Long Noncoding RNA, AFAP1-AS1, in Barrett's Esophagus and Esophageal Adenocarcinoma. Gastroenterology, 2013, 144, 956-966.e4.	1.3	216
63	Germline Inactivation of PTEN and Dysregulation of the Phosphoinositol-3-Kinase/Akt Pathway Cause Human Lhermitte-Duclos Disease in Adults. American Journal of Human Genetics, 2003, 73, 1191-1198.	6.2	213
64	A Meta-Analysis of Gaze Differences to Social and Nonsocial Information Between Individuals With and Without Autism. Journal of the American Academy of Child and Adolescent Psychiatry, 2017, 56, 546-555.	0.5	211
65	Association of germline mutation in the PTEN tumour suppressor gene and Proteus and Proteus-like syndromes. Lancet, The, 2001, 358, 210-211.	13.7	210
66	Somatic mutations in the RET proto-oncogene in sporadic medullary thyroid carcinoma. Clinical Endocrinology, 1996, 44, 249-257.	2.4	209
67	Somatic and occult germ-line mutations in SDHD, a mitochondrial complex II gene, in nonfamilial pheochromocytoma. Cancer Research, 2000, 60, 6822-5.	0.9	206
68	Analysis of PTEN and the 10q23 region in primary prostate carcinomas. Oncogene, 1998, 16, 1743-1748.	5.9	205
69	Germline Mutations and Variants in the Succinate Dehydrogenase Genes in Cowden and Cowden-like Syndromes. American Journal of Human Genetics, 2008, 83, 261-268.	6.2	205
70	Hamartomatous polyposis syndromes. Nature Reviews Gastroenterology & Hepatology, 2007, 4, 492-502.	1.7	204
71	Germline mutations of the RET ligand GDNF are not sufficient to cause Hirschsprung disease. Nature Genetics, 1996, 14, 345-347.	21.4	203
72	PTEN induces apoptosis and cell cycle arrest through phosphoinositol-3-kinase/Akt-dependent and -independent pathways. Human Molecular Genetics, 2001, 10, 237-242.	2.9	202

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73	Mapping Geographic Zones of Cancer Risk with Epigenetic Biomarkers in Normal Breast Tissue. Clinical Cancer Research, 2006, 12, 6626-6636.	7.0	201
74	Target identification among known drugs by deep learning from heterogeneous networks. Chemical Science, 2020, 11, 1775-1797.	7.4	193
75	Germline and germline mosaic PTEN mutations associated with a Proteus-like syndrome of hemihypertrophy, lower limb asymmetry, arteriovenous malformations and lipomatosis. Human Molecular Genetics, 2000, 9, 765-768.	2.9	188
76	Mutations of theRET proto-oncogene in the multiple endocrine neoplasia type 2 syndromes, related sporadic tumours, and Hirschsprung disease. Human Mutation, 1997, 9, 97-109.	2.5	180
77	Clinical Predictors for Germline Mutations in Head and Neck Paraganglioma Patients: Cost Reduction Strategy in Genetic Diagnostic Process as Fall-Out. Cancer Research, 2009, 69, 3650-3656.	0.9	178
78	PTEN suppresses breast cancer cell growth by phosphatase activity-dependent G1 arrest followed by cell death. Cancer Research, 1999, 59, 5808-14.	0.9	178
79	PTEN inhibits insulin-stimulated MEK/MAPK activation and cell growth by blocking IRS-1 phosphorylation and IRS-1/Grb-2/Sos complex formation in a breast cancer model. Human Molecular Genetics, 2001, 10, 605-616.	2.9	177
80	Nuclear-Cytoplasmic Partitioning of Phosphatase and Tensin Homologue Deleted on Chromosome 10 (PTEN) Differentially Regulates the Cell Cycle and Apoptosis. Cancer Research, 2005, 65, 8096-8100.	0.9	177
81	Germline PIK3CA and AKT1 Mutations in Cowden and Cowden-like Syndromes. American Journal of Human Genetics, 2013, 92, 76-80.	6.2	174
82	PTEN Mutational Spectra, Expression Levels, and Subcellular Localization in Microsatellite Stable and Unstable Colorectal Cancers. American Journal of Pathology, 2002, 161, 439-447.	3.8	173
83	Incidence and Clinical Characteristics of Thyroid Cancer in Prospective Series of Individuals with Cowden and Cowden-Like Syndrome Characterized by Germline PTEN, SDH, or KLLN Alterations. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E2063-E2071.	3.6	172
84	A novel point mutation in the tyrosine kinase domain of the RET proto-oncogene in sporadic medullary thyroid carcinoma and in a family with FMTC. Oncogene, 1995, 10, 509-13.	5.9	172
85	Low frequency of germline mutations in the RET proto-oncogene in patients with apparently sporadic medullary thyroid carcinoma. Clinical Endocrinology, 1995, 43, 123-127.	2.4	171
86	Nuclear PTEN expression and clinicopathologic features in a population-based series of primary cutaneous melanoma. International Journal of Cancer, 2002, 99, 63-67.	5.1	162
87	Multiple endocrine neoplasia type 2: An overview. Genetics in Medicine, 2011, 13, 755-764.	2.4	161
88	Mutation of theRET protooncogene in sporadic medullary thyroid carcinoma. Genes Chromosomes and Cancer, 1995, 12, 209-212.	2.8	160
89	Somatic mitochondrial DNA (mtDNA) mutations in papillary thyroid carcinomas and differential mtDNA sequence variants in cases with thyroid tumours. Oncogene, 2000, 19, 2060-2066.	5.9	160
90	Male breast cancer in Cowden syndrome patients with germline PTEN mutations. Journal of Medical Genetics, 2001, 38, 159-164.	3.2	160

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91	Clinical Predictors and Algorithm for the Genetic Diagnosis of Pheochromocytoma Patients. Clinical Cancer Research, 2009, 15, 6378-6385.	7.0	160
92	Bilateral Oophorectomy and Breast Cancer Risk inBRCA1andBRCA2Mutation Carriers. Journal of the National Cancer Institute, 2017, 109, .	6.3	160
93	Genetic model of multi-step breast carcinogenesis involving the epithelium and stroma: clues to tumour-microenvironment interactions. Human Molecular Genetics, 2001, 10, 1907-1913.	2.9	155
94	The pressure rises: update on the genetics of phaeochromocytoma. Human Molecular Genetics, 2002, 11, 2347-2354.	2.9	154
95	Genetic Classification of Benign and Malignant Thyroid Follicular Neoplasia Based on a Three-Gene Combination. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 2512-2521.	3.6	152
96	Familial Risks for Nonmedullary Thyroid Cancer. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 5747-5753.	3.6	151
97	Clinical Characterization of the Pheochromocytoma and Paraganglioma Susceptibility Genes SDHA, TMEM127, MAX, and SDHAF2 for Gene-Informed Prevention. JAMA Oncology, 2017, 3, 1204.	7.1	149
98	Implementation of Universal Microsatellite Instability and Immunohistochemistry Screening for Diagnosing Lynch Syndrome in a Large Academic Medical Center. Journal of Clinical Oncology, 2013, 31, 1336-1340.	1.6	147
99	Mutations in the RET proto-oncogene and the von Hippel-Lindau disease tumour suppressor gene in sporadic and syndromic phaeochromocytomas.. Journal of Medical Genetics, 1995, 32, 934-937.	3.2	144
100	Combined Total Genome Loss of Heterozygosity Scan of Breast Cancer Stroma and Epithelium Reveals Multiplicity of Stromal Targets. Cancer Research, 2004, 64, 7231-7236.	0.9	143
101	Mutations in DPC4 (SMAD4) cause juvenile polyposis syndrome, but only account for a minority of cases. Human Molecular Genetics, 1998, 7, 1907-1912.	2.9	142
102	Specific polymorphisms in the RETproto-oncogene are over-represented in patients with Hirschsprung disease and may represent loci modifying phenotypic expression. Journal of Medical Genetics, 1999, 36, 771-774.	3.2	142
103	Phosphatase and Tensin Homologue Deleted on Chromosome 10 (PTEN) Has Nuclear Localization Signalâ€“Like Sequences for Nuclear Import Mediated by Major Vault Protein. Cancer Research, 2005, 65, 4108-4116.	0.9	142
104	Genetics of Carney Triad: Recurrent Losses at Chromosome 1 but Lack of Germline Mutations in Genes Associated with Paragangliomas and Gastrointestinal Stromal Tumors. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2938-2943.	3.6	141
105	Germline PTEN mutations in Cowden syndrome-like families.. Journal of Medical Genetics, 1998, 35, 881-885.	3.2	140
106	A network medicine approach to investigation and population-based validation of disease manifestations and drug repurposing for COVID-19. PLoS Biology, 2020, 18, e3000970.	5.6	139
107	Germline Epigenetic Regulation of <emph type="ital">KILLIN</emph> in Cowden and Cowden-like Syndrome. JAMA - Journal of the American Medical Association, 2010, 304, 2724.	7.4	138
108	Over-representation of a germline RET sequence variant in patients with sporadic medullary thyroid carcinoma and somatic RET codon 918 mutation. Oncogene, 1999, 18, 1369-1373.	5.9	137

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109	Papillary and Follicular Thyroid Carcinomas Show Distinctly Different Microarray Expression Profiles and Can Be Distinguished by a Minimum of Five Genes. Journal of Clinical Oncology, 2004, 22, 3531-3539.	1.6	137
110	Outcomes of adrenal-sparing surgery or total adrenalectomy in phaeochromocytoma associated with multiple endocrine neoplasia type 2: an international retrospective population-based study. Lancet Oncology, The, 2014, 15, 648-655.	10.7	137
111	PTEN mutations are common in sporadic microsatellite stable colorectal cancer. Oncogene, 2004, 23, 617-628.	5.9	135
112	Cowden Syndrome. Seminars in Oncology, 2007, 34, 428-434.	2.2	135
113	A Novel Germline Mutation in BAP1 Predisposes to Familial Clear-Cell Renal Cell Carcinoma. Molecular Cancer Research, 2013, 11, 1061-1071.	3.4	135
114	A genome-wide positioning systems network algorithm for in silico drug repurposing. Nature Communications, 2019, 10, 3476.	12.8	134
115	The Clinical Spectrum of PTEN Mutations. Annual Review of Medicine, 2020, 71, 103-116.	12.2	134
116	Breast tissue, oral and urinary microbiomes in breast cancer. Oncotarget, 2017, 8, 88122-88138.	1.8	134
117	Interpreting epidemiological research: blinded comparison of methods used to estimate the prevalence of inherited mutations in BRCA1. Journal of Medical Genetics, 2001, 38, 824-833.	3.2	132
118	Molecular and phenotypic abnormalities in individuals with germline heterozygous PTEN mutations and autism. Molecular Psychiatry, 2015, 20, 1132-1138.	7.9	132
119	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. Journal of Clinical Oncology, 2016, 34, 2172-2181.	1.6	132
120	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	2.5	132
121	Genetic and phenotypic heterogeneity in the PTEN hamartoma tumour syndrome. Oncogene, 2008, 27, 5387-5397.	5.9	131
122	The Role of MMAC1 Mutations in Early-Onset Breast Cancer: Causative in Association with Cowden Syndrome and Excluded in BRCA1-Negative Cases. American Journal of Human Genetics, 1997, 61, 1036-1043.	6.2	128
123	PTEN-opathies: from biological insights to evidence-based precision medicine. Journal of Clinical Investigation, 2019, 129, 452-464.	8.2	128
124	RASSF1A promoter region CpG island hypermethylation in phaeochromocytomas and neuroblastoma tumours. Oncogene, 2001, 20, 7573-7577.	5.9	127
125	Contiguous Gene Deletion within Chromosome Arm 10q Is Associated with Juvenile Polyposis of Infancy, Reflecting Cooperation between the BMPR1A and PTEN Tumor-Suppressor Genes. American Journal of Human Genetics, 2006, 78, 1066-1074.	6.2	127
126	Gene Expression Profiling Identifies MMP-12 and ADAMDEC1 as Potential Pathogenic Mediators of Pulmonary Sarcoidosis. American Journal of Respiratory and Critical Care Medicine, 2009, 179, 929-938.	5.6	127

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127	GermlineNF1Mutational Spectra and Loss-of-Heterozygosity Analyses in Patients with Pheochromocytoma and Neurofibromatosis Type 1. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 2784-2792.	3.6	126
128	Frequent epigenetic inactivation of the SLIT2 gene in gliomas. Oncogene, 2003, 22, 4611-4616.	5.9	125
129	Geneâ€specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel. Human Mutation, 2018, 39, 1581-1592.	2.5	123
130	PTEN/MMAC1/TEP1 involvement in primary prostate cancers. Oncogene, 1998, 16, 2879-2883.	5.9	122
131	Pituitary Adenoma With Paraganglioma/Pheochromocytoma (3PAs) and Succinate Dehydrogenase Defects in Humans and Mice. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E710-E719.	3.6	121
132	Heterogeneous mutation of the RET proto-oncogene in subpopulations of medullary thyroid carcinoma. Cancer Research, 1996, 56, 2167-70.	0.9	120
133	Analysis of the 10q23 chromosomal region and the PTEN gene in human sporadic breast carcinoma. British Journal of Cancer, 1999, 79, 718-723.	6.4	119
134	Interpretation of Genetic Test Results for Hereditary Nonpolyposis Colorectal Cancer. JAMA - Journal of the American Medical Association, 1999, 282, 247.	7.4	118
135	Analysis of genetic and phenotypic heterogeneity in juvenile polyposis. Gut, 2000, 46, 656-660.	12.1	117
136	Risk profiles and penetrance estimations in multiple endocrine neoplasia type 2A caused by germline RET mutations located in exon 10. Human Mutation, 2011, 32, 51-58.	2.5	117
137	Loss of Expression of Protein Kinase A Regulatory Subunit 1Î± in Pigmented Epithelioid Melanocytoma But Not in Melanoma or Other Melanocytic Lesions. American Journal of Surgical Pathology, 2007, 31, 1764-1775.	3.7	115
138	Defining Phenotypes and Cancer Risk in Hyperplastic Polyposis Syndrome. Diseases of the Colon and Rectum, 2011, 54, 164-170.	1.3	114
139	Head and Neck Paragangliomas in Von Hippel-Lindau Disease and Multiple Endocrine Neoplasia Type 2. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1938-1944.	3.6	112
140	When Overgrowth Bumps Into Cancer: The PTENâ€Opathies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 114-121.	1.6	112
141	High Frequency of Submicroscopic Hemizygous Deletion Is a Major Mechanism of Loss of Expression of PTEN in Uveal Melanoma. Journal of Clinical Oncology, 2006, 24, 288-295.	1.6	110
142	Comprehensive characterization of proteinâ€“protein interactions perturbed by disease mutations. Nature Genetics, 2021, 53, 342-353.	21.4	109
143	Increased PTEN expression due to transcriptional activation of PPARÎ³ by Lovastatin and Rosiglitazone. International Journal of Cancer, 2006, 118, 2390-2398.	5.1	108
144	The ERK1/2 pathway modulates nuclear PTEN-mediated cell cycle arrest by cyclin D1 transcriptional regulation. Human Molecular Genetics, 2006, 15, 2553-2559.	2.9	106

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145	Cowden syndrome: Recognizing and managing a notâ€soâ€rare hereditary cancer syndrome. Journal of Surgical Oncology, 2015, 111, 125-130.	1.7	106
146	The Urinary Microbiome Differs Significantly Between Patients With Chronic Prostatitis/Chronic Pelvic Pain Syndrome and Controls as Well as Between Patients With Different Clinical Phenotypes. Urology, 2016, 92, 26-32.	1.0	106
147	GermlinePTEN mutation in a family with Cowden syndrome and Bannayan-Riley-Ruvalcaba Syndrome. American Journal of Medical Genetics Part A, 1998, 80, 399-402.	2.4	105
148	Second Malignant Neoplasms in Patients With Cowden Syndrome With Underlying Germline PTEN Mutations. Journal of Clinical Oncology, 2014, 32, 1818-1824.	1.6	105
149	Familiality in Barrett's Esophagus, Adenocarcinoma of the Esophagus, and Adenocarcinoma of the Gastroesophageal Junction. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 1668-1673.	2.5	104
150	Resveratrol enhances the anti-tumor activity of the mTOR inhibitor rapamycin in multiple breast cancer cell lines mainly by suppressing rapamycin-induced AKT signaling. Cancer Letters, 2011, 301, 168-176.	7.2	104
151	Genetic Testing for Cancer Predisposition. Annual Review of Medicine, 2001, 52, 371-400.	12.2	103
152	BMP2 exposure results in decreased PTEN protein degradation and increased PTEN levels. Human Molecular Genetics, 2003, 12, 679-684.	2.9	103
153	Genetic predisposition to phaeochromocytoma: analysis of candidate genes GDNF, RET and VHL. Human Molecular Genetics, 1997, 6, 1051-1056.	2.9	102
154	Implementation of Clinical Pharmacogenomics within a Large Health System: From Electronic Health Record Decision Support to Consultation Services. Pharmacotherapy, 2016, 36, 940-948.	2.6	102
155	Cowden syndrome and Lhermitte-Duclos disease in a family: a single genetic syndrome with pleiotropy?. Journal of Medical Genetics, 1994, 31, 458-461.	3.2	101
156	Genomic Instability Within Tumor Stroma and Clinicopathological Characteristics of Sporadic Primary Invasive Breast Carcinoma. JAMA - Journal of the American Medical Association, 2007, 297, 2103.	7.4	101
157	A Multi-Institutional Phase II Study of the Efficacy and Tolerability of Lapatinib in Patients with Advanced Hepatocellular Carcinomas. Clinical Cancer Research, 2009, 15, 5895-5901.	7.0	101
158	International trends in the uptake of cancer risk reduction strategies in women with a BRCA1 or BRCA2 mutation. British Journal of Cancer, 2019, 121, 15-21.	6.4	101
159	Human breast microbiome correlates with prognostic features and immunological signatures in breast cancer. Genome Medicine, 2021, 13, 60.	8.2	101
160	A Founding Locus within the RET Proto-Oncogene May Account for a Large Proportion of Apparently Sporadic Hirschsprung Disease and a Subset of Cases of Sporadic Medullary Thyroid Carcinoma. American Journal of Human Genetics, 2003, 72, 88-100.	6.2	100
161	Microenvironmental Genomic Alterations and Clinicopathological Behavior in Head and Neck Squamous Cell Carcinoma. JAMA - Journal of the American Medical Association, 2007, 297, 187.	7.4	100
162	Cowden Syndrome. Journal of Genetic Counseling, 1997, 6, 181-192.	1.6	99

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