Sharon A Savage

List of Publications by Year
in descending order

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269	17,948	13068	16605
papers	citations	68 h-index	123 g-index

323	323	323	20956
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Osteosarcoma incidence and survival rates from 1973 to 2004. Cancer, 2009, 115, 1531-1543.	2.0	1,758
2	International osteosarcoma incidence patterns in children and adolescents, middle ages and elderly persons. International Journal of Cancer, 2009, 125, 229-234.	2.3	557
3	Detectable clonal mosaicism and its relationship to aging and cancer. Nature Genetics, 2012, 44, 651-658.	9.4	519
4	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	9.4	493
5	A Genome-wide Association Study of Lung Cancer Identifies a Region of Chromosome 5p15 Associated with Risk for Adenocarcinoma. American Journal of Human Genetics, 2009, 85, 679-691.	2.6	489
6	Cancer in dyskeratosis congenita. Blood, 2009, 113, 6549-6557.	0.6	413
7	The Association of Telomere Length and Cancer: a Meta-analysis. Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 1238-1250.	1.1	386
8	TINF2, a Component of the Shelterin Telomere Protection Complex, Is Mutated in Dyskeratosis Congenita. American Journal of Human Genetics, 2008, 82, 501-509.	2.6	368
9	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45.	3.2	358
10	Risks of first and subsequent cancers among TP53 mutation carriers in the National Cancer Institute Liâ€Fraumeni syndrome cohort. Cancer, 2016, 122, 3673-3681.	2.0	346
11	Germline and somatic genetics of osteosarcoma â€" connecting aetiology, biology and therapy. Nature Reviews Endocrinology, 2017, 13, 480-491.	4.3	319
12	Very short telomere length by flow fluorescence in situ hybridization identifies patients with dyskeratosis congenita. Blood, 2007, 110, 1439-1447.	0.6	296
13	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. Nature Genetics, 2014, 46, 482-486.	9.4	283
14	Malignancies and survival patterns in the National Cancer Institute inherited bone marrow failure syndromes cohort study. British Journal of Haematology, 2010, 150, 179-188.	1.2	272
15	Cancer in the National Cancer Institute inherited bone marrow failure syndrome cohort after fifteen years of follow-up. Haematologica, 2018, 103, 30-39.	1.7	236
16	Telomere shortening and loss of self-renewal in dyskeratosis congenita induced pluripotent stem cells. Nature, 2011, 474, 399-402.	13.7	220
17	The association between leukocyte telomere length and cigarette smoking, dietary and physical variables, and risk of prostate cancer. Aging Cell, 2009, 8, 405-413.	3.0	217
18	Disruption of telomerase trafficking by TCAB1 mutation causes dyskeratosis congenita. Genes and Development, 2011, 25, 11-16.	2.7	213

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19	Using Epidemiology and Genomics to Understand Osteosarcoma Etiology. Sarcoma, 2011, 2011, 1-13.	0.7	210
20	The genetics and clinical manifestations of telomere biology disorders. Genetics in Medicine, 2010, 12, 753-764.	1.1	204
21	Germline mutations of regulator of telomere elongation helicase 1, RTEL1, in Dyskeratosis congenita. Human Genetics, 2013, 132, 473-480.	1.8	198
22	Dyskeratosis Congenita. Hematology/Oncology Clinics of North America, 2009, 23, 215-231.	0.9	195
23	Telomere length is associated with disease severity and declines with age in dyskeratosis congenita. Haematologica, 2012, 97, 353-359.	1.7	194
24	Genome-wide association study identifies two susceptibility loci for osteosarcoma. Nature Genetics, 2013, 45, 799-803.	9.4	181
25	The Built Environment: Designing Communities to Promote Physical Activity in Children. Pediatrics, 2009, 123, 1591-1598.	1.0	164
26	Updates on the biology and management of dyskeratosis congenita and related telomere biology disorders. Expert Review of Hematology, 2013, 6, 327-337.	1.0	157
27	Analysis of Heritability and Shared Heritability Based on Genome-Wide Association Studies for Thirteen Cancer Types. Journal of the National Cancer Institute, 2015, 107, djv279.	3.0	152
28	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634.	3.4	148
29	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in Patients With Osteosarcoma. JAMA Oncology, 2020, 6, 724.	3.4	139
30	Tobacco Use: A Pediatric Disease. Pediatrics, 2009, 124, 1474-1487.	1.0	132
31	Disease-specific hematopoietic cell transplantation: nonmyeloablative conditioning regimen for dyskeratosis congenita. Bone Marrow Transplantation, 2011, 46, 98-104.	1.3	132
32	Toward a Drug Development Path That Targets Metastatic Progression in Osteosarcoma. Clinical Cancer Research, 2014, 20, 4200-4209.	3.2	127
33	An update on the biology and management of dyskeratosis congenita and related telomere biology disorders. Expert Review of Hematology, 2019, 12, 1037-1052.	1.0	120
34	Telomere length in blood, buccal cells, and fibroblasts from patients with inherited bone marrow failure syndromes. Aging, 2010, 2, 867-874.	1.4	120
35	H/ACA Small RNA Dysfunctions in Disease Reveal Key Roles for Noncoding RNA Modifications in Hematopoietic Stem Cell Differentiation. Cell Reports, 2013, 3, 1493-1502.	2.9	109
36	Germline TP53 Variants and Susceptibility to Osteosarcoma. Journal of the National Cancer Institute, 2015, 107, .	3.0	109

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37	Outcomes of Allogeneic Hematopoietic Cell Transplantation in Patients with Dyskeratosis Congenita. Biology of Blood and Marrow Transplantation, 2013, 19, 1238-1243.	2.0	108
38	Telomere Length in Peripheral Leukocyte DNA and Gastric Cancer Risk. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 3103-3109.	1.1	106
39	A Recessive Founder Mutation in Regulator of Telomere Elongation Helicase 1, RTEL1, Underlies Severe Immunodeficiency and Features of Hoyeraal Hreidarsson Syndrome. PLoS Genetics, 2013, 9, e1003695.	1.5	106
40	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. Genes and Development, 2014, 28, 2090-2102.	2.7	106
41	Unraveling the pathogenesis of Hoyeraalâ€“Hreidarsson syndrome, a complex telomere biology disorder. British Journal of Haematology, 2015, 170, 457-471.	1.2	105
42	Beginning at the ends: telomeres and human disease. F1000Research, 2018, 7, 524.	0.8	105
43	Characterization of Large Structural Genetic Mosaicism in Human Autosomes. American Journal of Human Genetics, 2015, 96, 487-497.	2.6	101
44	Height at diagnosis and birth-weight as risk factors for osteosarcoma. Cancer Causes and Control, 2011, 22, 899-908.	0.8	99
45	Variants in the VCAM1 gene and risk for symptomatic stroke in sickle cell disease. Blood, 2002, 100, 4303-4309.	0.6	97
46	Genome-Wide Association Study of Relative Telomere Length. PLoS ONE, 2011, 6, e19635.	1.1	97
47	The association of telomere length and genetic variation in telomere biology genesa. Human Mutation, 2010, 31, 1050-1058.	1.1	93
48	A role for heterochromatin protein 1Î³ at human telomeres. Genes and Development, 2011, 25, 1807-1819.	2.7	93
49	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. Clinical Cancer Research, 2017, 23, e23-e31.	3.2	93
50	Imputation and subset-based association analysis across different cancer types identifies multiple independent risk loci in the TERT-CLPTM1L region on chromosome 5p15.33. Human Molecular Genetics, 2014, 23, 6616-6633.	1.4	90
51	Response to androgen therapy in patients with dyskeratosis congenita. British Journal of Haematology, 2014, 165, 349-357.	1.2	89
52	The genomics of inherited bone marrow failure: from mechanism to the clinic. British Journal of Haematology, 2017, 177, 526-542.	1.2	89
53	A Genome-Wide Scan Identifies Variants in NFIB Associated with Metastasis in Patients with Osteosarcoma. Cancer Discovery, 2015, 5, 920-931.	7.7	88
54	Female chromosome X mosaicism is age-related and preferentially affects the inactivated X chromosome. Nature Communications, 2016, 7, 11843.	5.8	86

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55	Iodine Deficiency, Pollutant Chemicals, and the Thyroid: New Information on an Old Problem. Pediatrics, 2014, 133, 1163-1166.	1.0	82
56	Telomeres and the natural lifespan limit in humans. Aging, 2017, 9, 1130-1142.	1.4	82
57	Global Climate Change and Children's Health. Pediatrics, 2007, 120, e1359-e1367.	1.0	81
58	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	1.1	81
59	Epidemiologic evidence for a role of telomere dysfunction in cancer etiology. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2012, 730, 75-84.	0.4	79
60	Whole-exome sequencing and functional studies identify RPS29 as a novel gene mutated in multicase Diamond-Blackfan anemia families. Blood, 2014, 124, 24-32.	0.6	79
61	Analysis of Genes Critical for Growth Regulation Identifies Insulin-like Growth Factor 2 Receptor Variations with Possible Functional Significance as Risk Factors for Osteosarcoma. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 1667-1674.	1.1	78
62	Erythrocyte adenosine deaminase: diagnostic value for Diamondâ€Blackfan anaemia. British Journal of Haematology, 2013, 160, 547-554.	1.2	76
63	Drinking Water From Private Wells and Risks to Children. Pediatrics, 2009, 123, e1123-e1137.	1.0	74
64	Association Between Donor Leukocyte Telomere Length and Survival After Unrelated Allogeneic Hematopoietic Cell Transplantation for Severe Aplastic Anemia. JAMA - Journal of the American Medical Association, 2015, 313, 594.	3.8	73
65	Ocular and Orbital Manifestations of the Inherited Bone Marrow Failure Syndromes: Fanconi Anemia and Dyskeratosis Congenita. Ophthalmology, 2010, 117, 615-622.	2.5	72
66	Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. Journal of Medical Genetics, 2017, 54, 417-425.	1.5	71
67	Genetic variation in five genes important in telomere biology and risk for breast cancer. British Journal of Cancer, 2007, 97, 832-836.	2.9	70
68	A comprehensive candidate gene approach identifies genetic variation associated with osteosarcoma. BMC Cancer, 2011, 11, 209.	1.1	69
69	Genomic Characterization of the Inherited Bone Marrow Failure Syndromes. Seminars in Hematology, 2013, 50, 333-347.	1.8	69
70	Recommended Guidelines for Validation, Quality Control, and Reporting of TP53 Variants in Clinical Practice. Cancer Research, 2017, 77, 1250-1260.	0.4	68
71	Dyskeratosis congenita: The first NIH clinical research workshop. Pediatric Blood and Cancer, 2009, 53, 520-523.	0.8	66
72	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105.	1.1	66

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73	The role of telomere biology in bone marrow failure and other disorders. Mechanisms of Ageing and Development, 2008, 129, 35-47.	2.2	64
74	Squamous cell carcinomas in patients with Fanconi anemia and dyskeratosis congenita: A search for human papillomavirus. International Journal of Cancer, 2013, 133, 1513-1515.	2.3	63
75	Lifetime Pesticide Use and Telomere Shortening among Male Pesticide Applicators in the Agricultural Health Study. Environmental Health Perspectives, 2013, 121, 919-924.	2.8	63
76	Telomere length in inherited bone marrow failure syndromes. Haematologica, 2015, 100, 49-54.	1.7	63
77	Interleukin-8 Polymorphisms Are Not Associated with Gastric Cancer Risk in a Polish Population. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 589-591.	1.1	60
78	Human Telomeres and Telomere Biology Disorders. Progress in Molecular Biology and Translational Science, 2014, 125, 41-66.	0.9	60
79	Leukocyte telomere length in a population-based caseâ€“control study of ovarian cancer: a pilot study. Cancer Causes and Control, 2010, 21, 77-82.	0.8	59
80	Classical inherited bone marrow failure syndromes with high risk for myelodysplastic syndrome and acute myelogenous leukemia. Seminars in Hematology, 2017, 54, 105-114.	1.8	57
81	Lung transplantation for pulmonary fibrosis in dyskeratosis congenita: Case Report and systematic literature review. BMC Blood Disorders, 2011, 11, 3.	0.9	56
82	LINE-1 methylation is inherited in familial testicular cancer kindreds. BMC Medical Genetics, 2010, 11, 77.	2.1	55
83	Common genetic variants in the 9p21 region and their associations with multiple tumours. British Journal of Cancer, 2013, 108, 1378-1386.	2.9	55
84	Effect of pre-analytic variables on the reproducibility of qPCR relative telomere length measurement. PLoS ONE, 2017, 12, e0184098.	1.1	55
85	Analysis of the Li-Fraumeni Spectrum Based on an International Germline TP53 Variant Data Set. JAMA Oncology, 2021, 7, 1800.	3.4	55
86	Genetic regulation of OAS1 nonsense-mediated decay underlies association with COVID-19 hospitalization in patients of European and African ancestries. Nature Genetics, 2022, 54, 1103-1116.	9.4	54
87	The TP53 Database: transition from the International Agency for Research on Cancer to the US National Cancer Institute. Cell Death and Differentiation, 2022, 29, 1071-1073.	5.0	53
88	Genetic variation, nucleotide diversity, and linkage disequilibrium in seven telomere stability genes suggest that these genes may be under constraint. Human Mutation, 2005, 26, 343-350.	1.1	50
89	Germline mutations in Protection of Telomeres 1 in two families with Hodgkin lymphoma. British Journal of Haematology, 2018, 181, 372-377.	1.2	48
90	Comparison of Chromosome Breakage in Non-Mosaic and Mosaic Patients with Fanconi Anemia, Relatives, and Patients with Other Inherited Bone Marrow Failure Syndromes. Cytogenetic and Genome Research, 2014, 144, 15-27.	0.6	47

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91	Genome-Wide Association Study Identifies Variants in Casein Kinase II (CSNK2A2) to be Associated With Leukocyte Telomere Length in a Punjabi Sikh Diabetic Cohort. Circulation: Cardiovascular Genetics, 2014, 7, 287-295.	5.1	46
92	Germ-line genetic variation ofTP53 in osteosarcoma. Pediatric Blood and Cancer, 2007, 49, 28-33.	0.8	45
93	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	3.7	45
94	Variants of the IL8 and IL8RB genes and risk for gastric cardia adenocarcinoma and esophageal squamous cell carcinoma. Cancer Epidemiology Biomarkers and Prevention, 2004, 13, 2251-7.	1.1	45
95	Determination of atrazine residues in food by enzyme immunoassay. Bulletin of Environmental Contamination and Toxicology, 1989, 42, 899-904.	1.3	44
96	Telomere Length and the Risk of Cutaneous Malignant Melanoma in Melanoma-Prone Families with and without CDKN2A Mutations. PLoS ONE, 2013, 8, e71121.	1.1	44
97	Late Effects Screening Guidelines after Hematopoietic Cell Transplantation for Inherited Bone Marrow Failure Syndromes: Consensus Statement From the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects After Pediatric HCT. Biology of Blood and Marrow Transplantation, 2017, 23, 1422-1428.	2.0	43
98	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. JAMA Oncology, 2017, 3, 1640.	3.4	43
99	Whole exome sequencing reveals a C-terminal germline variant in CEBPA-associated acute myeloid leukemia: 45-year follow up of a large family. Haematologica, 2016, 101, 846-852.	1.7	42
100	Correlation of Leukocyte Telomere Length Measurement Methods in Patients with Dyskeratosis Congenita and in Their Unaffected Relatives. International Journal of Molecular Sciences, 2017, 18, 1765.	1.8	42
101	Myelodysplastic Syndrome, Acute Myeloid Leukemia, and Cancer Surveillance in Fanconi Anemia. Hematology/Oncology Clinics of North America, 2018, 32, 657-668.	0.9	42
102	Reâ€equilibration of imbalanced NAD metabolism ameliorates the impact of telomere dysfunction. EMBO Journal, 2020, 39, e103420.	3.5	42
103	Pesticide Use and Relative Leukocyte Telomere Length in the Agricultural Health Study. PLoS ONE, 2015, 10, e0133382.	1.1	42
104	Global Climate Change and Children's Health. Pediatrics, 2007, 120, 1149-1152.	1.0	41
105	Parent decisionâ€making around the genetic testing of children for germline TP53 mutations. Cancer, 2015, 121, 286-293.	2.0	41
106	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. European Respiratory Journal, 2017, 49, 1601640.	3.1	41
107	Genetic variation in telomeric repeat binding factors 1 and 2 in aplastic anemia. Experimental Hematology, 2006, 34, 664-671.	0.2	40
108	Higher-than-expected population prevalence of potentially pathogenic germlineTP53variants in individuals unselected for cancer history. Human Mutation, 2017, 38, 1723-1730.	1.1	40

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109	Advancing <scp>RAS/RASopathy</scp> therapies: An NCIâ€sponsored intramural and extramural collaboration for the study of <scp>RASopathies</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 866-876.	0.7	40
110	The shelterin complex and hematopoiesis. Journal of Clinical Investigation, 2016, 126, 1621-1629.	3.9	40
111	Inhibition of acetyl cholinesterase by solanaceous glycoalkaloids and alkaloids. American Potato Journal, 1987, 64, 409-413.	0.4	39
112	Telomere biology in hematopoiesis and stem cell transplantation. Blood Reviews, 2011, 25, 261-269.	2.8	39
113	Genetic Variants Related to Longer Telomere Length are Associated with Increased Risk of Renal Cell Carcinoma. European Urology, 2017, 72, 747-754.	0.9	39
114	Inhibiting mitochondrial respiration prevents cancer in a mouse model of Li-Fraumeni syndrome. Journal of Clinical Investigation, 2016, 127, 132-136.	3.9	39
115	Drinking Water From Private Wells and Risks to Children. Pediatrics, 2009, 123, 1599-1605.	1.0	38
116	Current state of pediatric sarcoma biology and opportunities for future discovery: A report from the sarcoma translational research workshop. Cancer Genetics, 2016, 209, 182-194.	0.2	38
117	The integral membrane protein, ponticulin, acts as a monomer in nucleating actin assembly.. Journal of Cell Biology, 1993, 120, 909-922.	2.3	37
118	Neonatal manifestations of inherited bone marrow failure syndromes. Seminars in Fetal and Neonatal Medicine, 2016, 21, 57-65.	1.1	37
119	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	4.7	37
120	Parathyroid tumor development involves deregulation of homeobox genes. Endocrine-Related Cancer, 2008, 15, 267-275.	1.6	34
121	Immune status of patients with inherited bone marrow failure syndromes. American Journal of Hematology, 2015, 90, 702-708.	2.0	34
122	Disease progression and clinical outcomes in telomere biology disorders. Blood, 2022, 139, 1807-1819.	0.6	34
123	Polymorphisms in interleukin -2, -6, and -10 are not associated with gastric cardia or esophageal cancer in a high-risk chinese population. Cancer Epidemiology Biomarkers and Prevention, 2004, 13, 1547-9.	1.1	34
124	The Biomarkers of Exposure and Effect in Agriculture (BEEA) Study: Rationale, Design, Methods, and Participant Characteristics. Journal of Toxicology and Environmental Health - Part A: Current Issues, 2015, 78, 1338-1347.	1.1	32
125	Optimization of Therapy for Severe Aplastic Anemia Based on Clinical, Biologic, and Treatment Response Parameters: Conclusions of an International Working Group on Severe Aplastic Anemia Convened by the Blood and Marrow Transplant Clinical Trials Network, March 2010. Biology of Blood and Marrow Transplantation, 2011, 17, 291-299.	2.0	31
126	Bone marrow skeletal stem/progenitor cell defects in dyskeratosis congenita and telomere biology disorders. Blood, 2015, 125, 793-802.	0.6	31

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127	Current Knowledge and Priorities for Future Research in Late Effects after Hematopoietic Cell Transplantation for Inherited Bone Marrow Failure Syndromes: Consensus Statement from the Second Pediatric Blood and Marrow Transplant Consortium International Conference on Late Effects after Pediatric Hematopoietic Cell Transplantation. Biology of Blood and Marrow Transplantation, 2017, 23, 726-735.	2.0	31
128	Genomeâ€wide association study identifies the GLDC/IL33 locus associated with survival of osteosarcoma patients. International Journal of Cancer, 2018, 142, 1594-1601.	2.3	31
129	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. Blood Advances, 2018, 2, 1243-1249.	2.5	30
130	RTEL1 influences the abundance and localization of TERRA RNA. Nature Communications, 2021, 12, 3016.	5.8	30
131	Nucleotide diversity and population differentiation of the Melanocortin 1 Receptor gene, MC1R. BMC Genetics, 2008, 9, 31.	2.7	29
132	Dubowitz Syndrome Is a Complex Comprised of Multiple, Genetically Distinct and Phenotypically Overlapping Disorders. PLoS ONE, 2014, 9, e98686.	1.1	29
133	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. Pediatric Neurology, 2016, 56, 62-68.e1.	1.0	29
134	Cancer incidence, patterns, and genotypeâ€“phenotype associations in individuals with pathogenic or likely pathogenic germline TP53 variants: an observational cohort study. Lancet Oncology, The, 2021, 22, 1787-1798.	5.1	29
135	The relationship between DNA methylation and telomere length in dyskeratosis congenita. Aging Cell, 2012, 11, 24-28.	3.0	28
136	Telomere length and risk of glioma. Cancer Epidemiology, 2013, 37, 935-938.	0.8	28
137	Determination of methyl 2-benzimidazolecarbamate in fruit juices by immunoassay. Food Chemistry, 1990, 35, 51-58.	4.2	27
138	The international diffuse intrinsic pontine glioma registry: an infrastructure to accelerate collaborative research for an orphan disease. Journal of Neuro-Oncology, 2017, 132, 323-331.	1.4	27
139	Telomere length and variation in telomere biology genes in individuals with osteosarcoma. International Journal of Molecular Epidemiology and Genetics, 2011, 2, 19-29.	0.4	27
140	The accumulation and not the specific activity of telomerase ribonucleoprotein determines telomere maintenance deficiency in X-linked dyskeratosis congenita. Human Molecular Genetics, 2012, 21, 721-729.	1.4	26
141	Neuropsychiatric Conditions Among Patients with Dyskeratosis Congenita: A Link with Telomere Biology?. Psychosomatics, 2012, 53, 230-235.	2.5	26
142	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. Blood Cells, Molecules, and Diseases, 2006, 37, 134-136.	0.6	25
143	The Long and Short of Telomeres and Cancer Association Studies. Journal of the National Cancer Institute, 2013, 105, 448-449.	3.0	25
144	Novel FANCI mutations in Fanconi anemia with VACTERL association. American Journal of Medical Genetics, Part A, 2016, 170, 386-391.	0.7	25

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145	Genome-wide Association Study Identifies HLA-DPB1 as a Significant Risk Factor for Severe Aplastic Anemia. American Journal of Human Genetics, 2020, 106, 264-271.	2.6	25
146	Germline mutation of MDM4 , a major p53 regulator, in a familial syndrome of defective telomere maintenance. Science Advances, 2020, 6, eaay3511.	4.7	25
147	Research participant interest in primary, secondary, and incidental genomic findings. Genetics in Medicine, 2016, 18, 1218-1225.	1.1	24
148	Easing the Burden: Describing the Role of Social, Emotional and Spiritual Support in Research Families with Liâ€Fraumeni Syndrome. Journal of Genetic Counseling, 2016, 25, 529-542.	0.9	24
149	Mouse Homolog of the Human TP53 R337H Mutation Reveals Its Role in Tumorigenesis. Cancer Research, 2018, 78, 5375-5383.	0.4	24
150	Promoter methylation of candidate genes associated with familial testicular cancer. International Journal of Molecular Epidemiology and Genetics, 2012, 3, 213-27.	0.4	24
151	Connecting complex disorders through biology. Nature Genetics, 2012, 44, 238-240.	9.4	23
152	Telomeres in Molecular Epidemiology Studies. Progress in Molecular Biology and Translational Science, 2014, 125, 113-131.	0.9	23
153	Beyond the triad: Inheritance, mucocutaneous phenotype, and mortality in a cohort of patients with dyskeratosis congenita. Journal of the American Academy of Dermatology, 2018, 78, 804-806.	0.6	23
154	Telomere Length Calibration from qPCR Measurement: Limitations of Current Method. Cells, 2018, 7, 183.	1.8	23
155	Fundamental immuneâ€“oncogenicity trade-offs define driver mutationÂfitness. Nature, 2022, 606, 172-179.	13.7	23
156	Structure and function of the human Gly1619Arg polymorphism of M6P/IGF2R domain 11 implicated in IGF2 dependent growth. Journal of Molecular Endocrinology, 2009, 42, 341-356.	1.1	22
157	Genetic variation at chromosome 8q24 in osteosarcoma cases and controls. Carcinogenesis, 2010, 31, 1400-1404.	1.3	22
158	Investigation of chromosome X inactivation and clinical phenotypes in female carriers of DKC1 mutations. American Journal of Hematology, 2016, 91, 1215-1220.	2.0	22
159	Effect of Recipient Age and Stem Cell Source on the Association between Donor Telomere Length and Survival after Allogeneic Unrelated Hematopoietic Cell Transplantation for Severe Aplastic Anemia. Biology of Blood and Marrow Transplantation, 2016, 22, 2276-2282.	2.0	22
160	Penetrance of Different Cancer Types in Families with Li-Fraumeni Syndrome: A Validation Study Using Multicenter Cohorts. Cancer Research, 2020, 80, 354-360.	0.4	22
161	Case-parent analysis of variation in pubertal hormone genes and pediatric osteosarcoma: a Children's Oncology Group (COG) study. International Journal of Molecular Epidemiology and Genetics, 2012, 3, 286-93.	0.4	22
162	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. British Journal of Haematology, 2013, 162, 542-546.	1.2	21

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163	Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. Journal of Psychosocial Oncology, 2019, 37, 178-193.	0.6	21
164	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. Molecular Genetics & Genomic Medicine, 2016, 4, 475-479.	0.6	20
165	Alternative splicing is a developmental switch for hTERT expression. Molecular Cell, 2021, 81, 2349-2360.e6.	4.5	19
166	Carrier screening of RTEL1 mutations in the Ashkenazi Jewish population. Clinical Genetics, 2015, 88, 177-181.	1.0	18
167	On the Interplay of Telomeres, Nevi and the Risk of Melanoma. PLoS ONE, 2012, 7, e52466.	1.1	18
168	Telomere length and epigenetic clocks as markers of cellular aging: a comparative study. GeroScience, 2022, 44, 1861-1869.	2.1	18
169	Genetic testing in severe aplastic anemia is required forÂoptimal hematopoietic cell transplant outcomes. Blood, 2022, 140, 909-921.	0.6	18
170	Risk of renal cell carcinoma in relation to blood telomere length in a population-based caseâ€“control study. British Journal of Cancer, 2011, 105, 1772-1775.	2.9	17
171	CNS manifestations in patients with telomere biology disorders. Neurology: Genetics, 2019, 5, 370.	0.9	17
172	Catheter-directed thrombolysis in a child with acute lymphoblastic leukemia and extensive deep vein thrombosis. , 2000, 34, 215-217.		16
173	Patterns of Bone Sarcomas as a Second Malignancy in Relation to Radiotherapy in Adulthood and Histologic Type. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1993-1999.	1.1	16
174	Relationship between plasma 25-hydroxyvitamin D and leucocyte telomere length by sex and race in a US study. British Journal of Nutrition, 2016, 116, 953-960.	1.2	16
175	Is the osteosarcoma genome targetable?. Nature Reviews Endocrinology, 2017, 13, 506-508.	4.3	16
176	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. BMC Medical Genetics, 2018, 19, 85.	2.1	16
177	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562.	1.1	16
178	Pregnancies in patients with inherited bone marrow failure syndromes in the NCI cohort. Blood, 2017, 130, 1674-1676.	0.6	15
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