Sharon A Savage

List of Publications by Citations

253 papers	13,498 citations	59 h-index	110 g-index
323 ext. papers	15,831 ext. citations	6.7 avg, IF	6.59 L-index

#	Paper	IF	Citations
253	Osteosarcoma incidence and survival rates from 1973 to 2004: data from the Surveillance, Epidemiology, and End Results Program. *Cancer*, **2009**, 115, 1531-43	6.4	1385
252	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, 679-91	11	442
251	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
250	Detectable clonal mosaicism and its relationship to aging and cancer. *Nature Genetics*, **2012**, 44, 651-8	36.3	409
249	International osteosarcoma incidence patterns in children and adolescents, middle ages and elderly persons. *International Journal of Cancer*, **2009**, 125, 229-34	7.5	399
248	Cancer in dyskeratosis congenita. *Blood*, **2009**, 113, 6549-57	2.2	353
247	The association of telomere length and cancer: a meta-analysis. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 1238-50	4	349
246	TINF2, a component of the shelterin telomere protection complex, is mutated in dyskeratosis congenita. *American Journal of Human Genetics*, **2008**, 82, 501-9	11	312
245	Very short telomere length by flow fluorescence in situ hybridization identifies patients with dyskeratosis congenita. *Blood*, **2007**, 110, 1439-47	2.2	267
244	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. *Clinical Cancer Research*, **2017**, 23, e38-e45	12.9	245
243	Risks of first and subsequent cancers among TP53 mutation carriers in the National Cancer Institute Li-Fraumeni syndrome cohort. *Cancer*, **2016**, 122, 3673-3681	6.4	236
242	Germline and somatic genetics of osteosarcoma - connecting aetiology, biology and therapy. *Nature Reviews Endocrinology*, **2017**, 13, 480-491	15.2	225
241	Malignancies and survival patterns in the National Cancer Institute inherited bone marrow failure syndromes cohort study. *British Journal of Haematology*, **2010**, 150, 179-88	4.5	224
240	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. *Nature Genetics*, **2014**, 46, 482-6	36.3	220
239	Telomere shortening and loss of self-renewal in dyskeratosis congenita induced pluripotent stem cells. *Nature*, **2011**, 474, 399-402	50.4	186
238	The association between leukocyte telomere length and cigarette smoking, dietary and physical variables, and risk of prostate cancer. *Aging Cell*, **2009**, 8, 405-13	9.9	186
237	Disruption of telomerase trafficking by TCAB1 mutation causes dyskeratosis congenita. *Genes and Development*, **2011**, 25, 11-6	12.6	178

236	Germline mutations of regulator of telomere elongation helicase 1, RTEL1, in Dyskeratosis congenita. *Human Genetics*, **2013**, 132, 473-80	6.3	162
235	Using epidemiology and genomics to understand osteosarcoma etiology. *Sarcoma*, **2011**, 2011, 548151	3.1	159
234	The genetics and clinical manifestations of telomere biology disorders. *Genetics in Medicine*, **2010**, 12, 753-64	8.1	158
233	Genome-wide association study identifies two susceptibility loci for osteosarcoma. *Nature Genetics*, **2013**, 45, 799-803	36.3	156
232	Dyskeratosis congenita. *Hematology/Oncology Clinics of North America*, **2009**, 23, 215-31	3.1	151
231	Telomere length is associated with disease severity and declines with age in dyskeratosis congenita. *Haematologica*, **2012**, 97, 353-9	6.6	145
230	Cancer in the National Cancer Institute inherited bone marrow failure syndrome cohort after fifteen years of follow-up. *Haematologica*, **2018**, 103, 30-39	6.6	134
229	The built environment: designing communities to promote physical activity in children. *Pediatrics*, **2009**, 123, 1591-8	7.4	134
228	From the American Academy of Pediatrics: Policy statement--Tobacco use: a pediatric disease. *Pediatrics*, **2009**, 124, 1474-87	7.4	115
227	Disease-specific hematopoietic cell transplantation: nonmyeloablative conditioning regimen for dyskeratosis congenita. *Bone Marrow Transplantation*, **2011**, 46, 98-104	4.4	108
226	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging: A Meta-analysis. *JAMA Oncology*, **2017**, 3, 1634-1639	13.4	107
225	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	9.7	107
224	Toward a drug development path that targets metastatic progression in osteosarcoma. *Clinical Cancer Research*, **2014**, 20, 4200-9	12.9	103
223	Telomere length in blood, buccal cells, and fibroblasts from patients with inherited bone marrow failure syndromes. *Aging*, **2010**, 2, 867-74	5.6	103
222	Updates on the biology and management of dyskeratosis congenita and related telomere biology disorders. *Expert Review of Hematology*, **2013**, 6, 327-37	2.8	98
221	Telomere length in peripheral leukocyte DNA and gastric cancer risk. *Cancer Epidemiology Biomarkers and Prevention*, **2009**, 18, 3103-9	4	96
220	Outcomes of allogeneic hematopoietic cell transplantation in patients with dyskeratosis congenita. *Biology of Blood and Marrow Transplantation*, **2013**, 19, 1238-43	4.7	86
219	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. *Genes and Development*, **2014**, 28, 2090-102	12.6	86

218	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	6	85
217	Height at diagnosis and birth-weight as risk factors for osteosarcoma. *Cancer Causes and Control*, **2011**, 22, 899-908	2.8	84
216	Variants in the VCAM1 gene and risk for symptomatic stroke in sickle cell disease. *Blood*, **2002**, 100, 4303-9	22	83
215	The association of telomere length and genetic variation in telomere biology genes. *Human Mutation*, **2010**, 31, 1050-8	4.7	82
214	H/ACA small RNA dysfunctions in disease reveal key roles for noncoding RNA modifications in hematopoietic stem cell differentiation. *Cell Reports*, **2013**, 3, 1493-502	10.6	81
213	Genome-wide association study of relative telomere length. *PLoS ONE*, **2011**, 6, e19635	3.7	80
212	Unraveling the pathogenesis of Hoyeraal-Hreidarsson syndrome, a complex telomere biology disorder. *British Journal of Haematology*, **2015**, 170, 457-71	4.5	79
211	Characterization of large structural genetic mosaicism in human autosomes. *American Journal of Human Genetics*, **2015**, 96, 487-97	11	77
210	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, 6616-33	5.6	77
209	A role for heterochromatin protein 1 at human telomeres. *Genes and Development*, **2011**, 25, 1807-19	12.6	76
208	Germline TP53 variants and susceptibility to osteosarcoma. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	75
207	A Genome-Wide Scan Identifies Variants in NFIB Associated with Metastasis in Patients with Osteosarcoma. *Cancer Discovery*, **2015**, 5, 920-31	24.4	71
206	Beginning at the ends: telomeres and human disease. *F1000Research*, **2018**, 7,	3.6	71
205	Epidemiologic evidence for a role of telomere dysfunction in cancer etiology. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2012**, 730, 75-84	3.3	71
204	Whole-exome sequencing and functional studies identify RPS29 as a novel gene mutated in multicase Diamond-Blackfan anemia families. *Blood*, **2014**, 124, 24-32	2.2	69
203	Iodine deficiency, pollutant chemicals, and the thyroid: new information on an old problem. *Pediatrics*, **2014**, 133, 1163-6	7.4	68
202	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, 1667-74	4	68
201	Response to androgen therapy in patients with dyskeratosis congenita. *British Journal of Haematology*, **2014**, 165, 349-57	4.5	67

200	The genomics of inherited bone marrow failure: from mechanism to the clinic. *British Journal of Haematology*, **2017**, 177, 526-542	4.5	65
199	Genetic variation in five genes important in telomere biology and risk for breast cancer. *British Journal of Cancer*, **2007**, 97, 832-6	8.7	65
198	Ocular and orbital manifestations of the inherited bone marrow failure syndromes: Fanconi anemia and dyskeratosis congenita. *Ophthalmology*, **2010**, 117, 615-22	7.3	64
197	An update on the biology and management of dyskeratosis congenita and related telomere biology disorders. *Expert Review of Hematology*, **2019**, 12, 1037-1052	2.8	60
196	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in Patients With Osteosarcoma. *JAMA Oncology*, **2020**, 6, 724-734	13.4	60
195	Female chromosome X mosaicism is age-related and preferentially affects the inactivated X chromosome. *Nature Communications*, **2016**, 7, 11843	17.4	59
194	Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. *Journal of Medical Genetics*, **2017**, 54, 417-425	5.8	57
193	Genomic characterization of the inherited bone marrow failure syndromes. *Seminars in Hematology*, **2013**, 50, 333-47	4	57
192	Dyskeratosis congenita: the first NIH clinical research workshop. *Pediatric Blood and Cancer*, **2009**, 53, 520-3	3	57
191	The role of telomere biology in bone marrow failure and other disorders. *Mechanisms of Ageing and Development*, **2008**, 129, 35-47	5.6	57
190	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. *Clinical Cancer Research*, **2017**, 23, e23-e31	12.9	56
189	Squamous cell carcinomas in patients with Fanconi anemia and dyskeratosis congenita: a search for human papillomavirus. *International Journal of Cancer*, **2013**, 133, 1513-5	7.5	56
188	Erythrocyte adenosine deaminase: diagnostic value for Diamond-Blackfan anaemia. *British Journal of Haematology*, **2013**, 160, 547-54	4.5	56
187	Leukocyte telomere length in a population-based case-control study of ovarian cancer: a pilot study. *Cancer Causes and Control*, **2010**, 21, 77-82	2.8	55
186	Global climate change and children's health. *Pediatrics*, **2007**, 120, e1359-67	7.4	55
185	A comprehensive candidate gene approach identifies genetic variation associated with osteosarcoma. *BMC Cancer*, **2011**, 11, 209	4.8	54
184	Interleukin-8 polymorphisms are not associated with gastric cancer risk in a Polish population. *Cancer Epidemiology Biomarkers and Prevention*, **2006**, 15, 589-91	4	54
183	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, 594-602	27.4	53

182	Telomeres and the natural lifespan limit in humans. *Aging*, **2017**, 9, 1130-1142	5.6	53
181	Telomere length in inherited bone marrow failure syndromes. *Haematologica*, **2015**, 100, 49-54	6.6	52
180	Lifetime pesticide use and telomere shortening among male pesticide applicators in the Agricultural Health Study. *Environmental Health Perspectives*, **2013**, 121, 919-24	8.4	51
179	LINE-1 methylation is inherited in familial testicular cancer kindreds. *BMC Medical Genetics*, **2010**, 11, 77	2.1	51
178	Drinking water from private wells and risks to children. *Pediatrics*, **2009**, 123, e1123-37	7.4	50
177	Lung transplantation for pulmonary fibrosis in dyskeratosis congenita: Case Report and systematic literature review. *BMC Blood Disorders*, **2011**, 11, 3		49
176	Genetic variation, nucleotide diversity, and linkage disequilibrium in seven telomere stability genes suggest that these genes may be under constraint. *Human Mutation*, **2005**, 26, 343-50	4.7	49
175	Recommended Guidelines for Validation, Quality Control, and Reporting of Variants in Clinical Practice. *Cancer Research*, **2017**, 77, 1250-1260	10.1	45
174	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	4	45
173	Human telomeres and telomere biology disorders. *Progress in Molecular Biology and Translational Science*, **2014**, 125, 41-66	4	44
172	Classical inherited bone marrow failure syndromes with high risk for myelodysplastic syndrome and acute myelogenous leukemia. *Seminars in Hematology*, **2017**, 54, 105-114	4	40
171	Common genetic variants in the 9p21 region and their associations with multiple tumours. *British Journal of Cancer*, **2013**, 108, 1378-86	8.7	40
170	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. *Human Mutation*, **2019**, 40, 97-105	4.7	39
169	Telomere length and the risk of cutaneous malignant melanoma in melanoma-prone families with and without CDKN2A mutations. *PLoS ONE*, **2013**, 8, e71121	3.7	37
168	Germ-line genetic variation of TP53 in osteosarcoma. *Pediatric Blood and Cancer*, **2007**, 49, 28-33	3	37
167	Genetic variation in telomeric repeat binding factors 1 and 2 in aplastic anemia. *Experimental Hematology*, **2006**, 34, 664-71	3.1	37
166	Global climate change and children's health. *Pediatrics*, **2007**, 120, 1149-52	7.4	37
165	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, 287-95		36

164	Parent decision-making around the genetic testing of children for germline TP53 mutations. *Cancer* , **2015**, 121, 286-93	6.4	35
163	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. *JAMA Oncology*, **2017**, 3, 1640-1645	13.4	35
162	Determination of atrazine residues in food by enzyme immunoassay. *Bulletin of Environmental Contamination and Toxicology*, **1989**, 42, 899-904	2.7	35
161	The integral membrane protein, ponticulin, acts as a monomer in nucleating actin assembly. *Journal of Cell Biology*, **1993**, 120, 909-22	7.3	34
160	Inhibition of acetyl cholinesterase by solanaceous glycoalkaloids and alkaloids. *American Potato Journal*, **1987**, 64, 409-413		34
159	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	4	34
158	Telomere biology in hematopoiesis and stem cell transplantation. *Blood Reviews*, **2011**, 25, 261-9	11.1	32
157	Parathyroid tumor development involves deregulation of homeobox genes. *Endocrine-Related Cancer*, **2008**, 15, 267-75	5.7	31
156	Pesticide Use and Relative Leukocyte Telomere Length in the Agricultural Health Study. *PLoS ONE*, **2015**, 10, e0133382	3.7	31
155	The shelterin complex and hematopoiesis. *Journal of Clinical Investigation*, **2016**, 126, 1621-9	15.9	30
154	Effect of pre-analytic variables on the reproducibility of qPCR relative telomere length measurement. *PLoS ONE*, **2017**, 12, e0184098	3.7	30
153	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. *European Respiratory Journal*, **2017**, 49,	13.6	29
152	Drinking water from private wells and risks to children. *Pediatrics*, **2009**, 123, 1599-605	7.4	29
151	Current state of pediatric sarcoma biology and opportunities for future discovery: A report from the sarcoma translational research workshop. *Cancer Genetics*, **2016**, 209, 182-94	2.3	29
150	Understanding the evolving phenotype of vascular complications in telomere biology disorders. *Angiogenesis*, **2019**, 22, 95-102	10.6	29
149	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. *Human Mutation*, **2021**, 42, 223-236	4.7	29
148	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	4.7	28
147	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	1.9	28

146	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, 291-9	4.7	28
145	Genetic Variants Related to Longer Telomere Length are Associated with Increased Risk of Renal Cell Carcinoma. *European Urology*, **2017**, 72, 747-754	10.2	27
144	Nucleotide diversity and population differentiation of the melanocortin 1 receptor gene, MC1R. *BMC Genetics*, **2008**, 9, 31	2.6	27
143	Telomere length and risk of glioma. *Cancer Epidemiology*, **2013**, 37, 935-8	2.8	26
142	Bone marrow skeletal stem/progenitor cell defects in dyskeratosis congenita and telomere biology disorders. *Blood*, **2015**, 125, 793-802	2.2	26
141	Dubowitz syndrome is a complex comprised of multiple, genetically distinct and phenotypically overlapping disorders. *PLoS ONE*, **2014**, 9, e98686	3.7	26
140	Inhibiting mitochondrial respiration prevents cancer in a mouse model of Li-Fraumeni syndrome. *Journal of Clinical Investigation*, **2017**, 127, 132-136	15.9	26
139	Telomere length and variation in telomere biology genes in individuals with osteosarcoma. *International Journal of Molecular Epidemiology and Genetics*, **2011**, 2, 19-29	0.9	26
138	Myelodysplastic Syndrome, Acute Myeloid Leukemia, and Cancer Surveillance in Fanconi Anemia. *Hematology/Oncology Clinics of North America*, **2018**, 32, 657-668	3.1	26
137	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*	4.7	25
136	Neonatal manifestations of inherited bone marrow failure syndromes. *Seminars in Fetal and Neonatal Medicine*, **2016**, 21, 57-65	3.7	25
135	Determination of methyl 2-benzimidazolecarbamate in fruit juices by immunoassay. *Food Chemistry* , **1990**, 35, 51-58	8.5	25
134	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, 846-52	6.6	25
133	Higher-than-expected population prevalence of potentially pathogenic germline TP53 variants in individuals unselected for cancer history. *Human Mutation*, **2017**, 38, 1723-1730	4.7	24
132	Advancing RAS/RASopathy therapies: An NCI-sponsored intramural and extramural collaboration for the study of RASopathies. *American Journal of Medical Genetics, Part A*, **2020**, 182, 866-876	2.5	24
131	Germline mutations in Protection of Telomeres 1 in two families with Hodgkin lymphoma. *British Journal of Haematology*, **2018**, 181, 372-377	4.5	23
130	Correlation of Leukocyte Telomere Length Measurement Methods in Patients with Dyskeratosis Congenita and in Their Unaffected Relatives. *International Journal of Molecular Sciences*, **2017**, 18,	6.3	23
129	The relationship between DNA methylation and telomere length in dyskeratosis congenita. *Aging Cell*, **2012**, 11, 24-8	9.9	23

128	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. *Pediatric Neurology*, **2016**, 56, 62-68.e1	2.9	23
127	Promoter methylation of candidate genes associated with familial testicular cancer. *International Journal of Molecular Epidemiology and Genetics*, **2012**, 3, 213-27	0.9	22
126	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, 1338-47	3.2	21
125	Novel FANCI mutations in Fanconi anemia with VACTERL association. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 386-391	2.5	20
124	Immune status of patients with inherited bone marrow failure syndromes. *American Journal of Hematology*, **2015**, 90, 702-8	7.1	20
123	Connecting complex disorders through biology. *Nature Genetics*, **2012**, 44, 238-40	36.3	20
122	The accumulation and not the specific activity of telomerase ribonucleoprotein determines telomere maintenance deficiency in X-linked dyskeratosis congenita. *Human Molecular Genetics*, **2012**, 21, 721-9	5.6	20
121	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. *Blood Cells, Molecules, and Diseases*, **2006**, 37, 134-6	2.1	20
120	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	4.8	19
119	Neuropsychiatric conditions among patients with dyskeratosis congenita: a link with telomere biology?. *Psychosomatics*, **2012**, 53, 230-5	2.6	19
118	Structure and function of the human Gly1619Arg polymorphism of M6P/IGF2R domain 11 implicated in IGF2 dependent growth. *Journal of Molecular Endocrinology*, **2009**, 42, 341-56	4.5	19
117	Case-parent analysis of variation in pubertal hormone genes and pediatric osteosarcoma: a Children Oncology Group (COG) study. *International Journal of Molecular Epidemiology and Genetics*, **2012**, 3, 286-93	0.9	19
116	Research participant interest in primary, secondary, and incidental genomic findings. *Genetics in Medicine*, **2016**, 18, 1218-1225	8.1	19
115	Genome-wide association study identifies the GLDC/IL33 locus associated with survival of osteosarcoma patients. *International Journal of Cancer*, **2018**, 142, 1594-1601	7.5	19
114	Investigation of chromosome X inactivation and clinical phenotypes in female carriers of DKC1 mutations. *American Journal of Hematology*, **2016**, 91, 1215-1220	7.1	18
113	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	4.7	18
112	Telomeres in molecular epidemiology studies. *Progress in Molecular Biology and Translational Science*, **2014**, 125, 113-31	4	18
111	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. *Blood Advances*, **2018**, 2, 1243-1249	7.8	18

110	Easing the Burden: Describing the Role of Social, Emotional and Spiritual Support in Research Families with Li-Fraumeni Syndrome. *Journal of Genetic Counseling*, **2016**, 25, 529-42	2.5	17
109	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. *British Journal of Haematology*, **2013**, 162, 542-6	4.5	17
108	Mouse Homolog of the Human R337H Mutation Reveals Its Role in Tumorigenesis. *Cancer Research*, **2018**, 78, 5375-5383	10.1	16
107	Carrier screening of RTEL1 mutations in the Ashkenazi Jewish population. *Clinical Genetics*, **2015**, 88, 177-81	4	16
106	Genetic variation at chromosome 8q24 in osteosarcoma cases and controls. *Carcinogenesis*, **2010**, 31, 1400-4	4.6	16
105	Risk of renal cell carcinoma in relation to blood telomere length in a population-based case-control study. *British Journal of Cancer*, **2011**, 105, 1772-5	8.7	16
104	Re-equilibration of imbalanced NAD metabolism ameliorates the impact of telomere dysfunction. *EMBO Journal*, **2020**, 39, e103420	13	16
103	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 475-9	2.3	16
102	On the interplay of telomeres, nevi and the risk of melanoma. *PLoS ONE*, **2012**, 7, e52466	3.7	15
101	XAF1 as a modifier of p53 function and cancer susceptibility. *Science Advances*, **2020**, 6, eaba3231	14.3	14
100	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	4.5	14
99	Catheter-directed thrombolysis in a child with acute lymphoblastic leukemia and extensive deep vein thrombosis. *Medical and Pediatric Oncology*, **2000**, 34, 215-7		14
98	Telomere Length Calibration from qPCR Measurement: Limitations of Current Method. *Cells*, **2018**, 7,	7.9	14
97	Patterns of bone sarcomas as a second malignancy in relation to radiotherapy in adulthood and histologic type. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 1993-9	4	13
96	Antibody response to human papillomavirus vaccine in subjects with inherited bone marrow failure syndromes. *Vaccine*, **2014**, 32, 1169-73	4.1	12
95	Reduced serum levels of anti-Müllerian hormone in females with inherited bone marrow failure syndromes. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E197-203	5.6	12
94	Relationship between plasma 25-hydroxyvitamin D and leucocyte telomere length by sex and race in a US study. *British Journal of Nutrition*, **2016**, 116, 953-60	3.6	12
93	No association between donor telomere length and outcomes after allogeneic unrelated hematopoietic cell transplant in patients with acute leukemia. *Bone Marrow Transplantation*, **2018**, 53, 383-391	4.4	11

92	Bone cancer: Is the osteosarcoma genome targetable?. *Nature Reviews Endocrinology*, **2017**, 13, 506-508	15.2	11
91	Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. *Journal of Psychosocial Oncology*, **2019**, 37, 178-193	2.8	10
90	Donor telomere length and causes of death after unrelated hematopoietic cell transplantation in patients with marrow failure. *Blood*, **2018**, 131, 2393-2398	2.2	10
89	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. *BMC Medical Genetics*, **2018**, 19, 85	2.1	10
88	Pregnancies in patients with inherited bone marrow failure syndromes in the NCI cohort. *Blood*, **2017**, 130, 1674-1676	2.2	10
87	Worldwide genetic structure in 37 genes important in telomere biology. *Heredity*, **2012**, 108, 124-33	3.6	10
86	Telomere stability genes are not mutated in osteosarcoma cell lines. *Cancer Genetics and Cytogenetics*, **2005**, 160, 79-81		10
85	RTEL1 influences the abundance and localization of TERRA RNA. *Nature Communications*, **2021**, 12, 3016	17.4	10
84	Suggested application of HER2+ breast tumor phenotype for germline TP53 variant classification within ACMG/AMP guidelines. *Human Mutation*, **2020**, 41, 1555-1562	4.7	9
83	Germline mutation of , a major p53 regulator, in a familial syndrome of defective telomere maintenance. *Science Advances*, **2020**, 6, eaay3511	14.3	9
82	CNS manifestations in patients with telomere biology disorders. *Neurology: Genetics*, **2019**, 5, 370	3.8	9
81	Using germ-line genetic variation to investigate and treat cancer. *Drug Discovery Today*, **2004**, 9, 610-8	8.8	9
80	Penetrance of Different Cancer Types in Families with Li-Fraumeni Syndrome: A Validation Study Using Multicenter Cohorts. *Cancer Research*, **2020**, 80, 354-360	10.1	9
79	Prognostic significance of pulmonary function tests in dyskeratosis congenita, a telomere biology disorder. *ERJ Open Research*, **2019**, 5,	3.5	9
78	Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. *Genome Medicine*, **2018**, 10, 99	14.4	9
77	Bone mineral density in patients with inherited bone marrow failure syndromes. *Pediatric Research*, **2017**, 82, 458-464	3.2	8
76	Relative Telomere Length before Hematopoietic Cell Transplantation and Outcome after Unrelated Donor Hematopoietic Cell Transplantation for Acute Leukemia. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1054-1058	4.7	8
75	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	11	8

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