Sharon A Savage

List of Publications by Year in Descending Order

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	The TP53 Database: transition from the International Agency for Research on Cancer to the US National Cancer Institute.. *Cell Death and Differentiation*, **2022**,	12.7	3
252	Fundamental immune-oncogenicity trade-offs define driver mutation fitness.. *Nature*, **2022**,	50.4	1
251	Lipoprotein particle alterations due to androgen therapy in individuals with dyskeratosis congenita.. *EBioMedicine*, **2021**, 75, 103760	8.8	1
250	Whole Exome Sequencing in Severe Aplastic Anemia Identifies Unrecognized Inherited Subset with Inferior Survival after Hematopoietic Cell Transplant. *Blood*, **2021**, 138, 605-605	2.2	
249	Genomic-Based Machine Learning Towards Prediction of the Etiology of Bone Marrow Failure Syndromes. *Blood*, **2021**, 138, 2182-2182	2.2	1
248	Germline-Somatic Interactions in Myelofibrosis Susceptibility. *Blood*, **2021**, 138, 313-313	2.2	
247	Cancer incidence, patterns, and genotype-phenotype associations in individuals with pathogenic or likely pathogenic germline TP53 variants: an observational cohort study. *Lancet Oncology, The*, **2021**, 22, 1787-1798	21.7	4
246	Disease Progression and Clinical Outcomes in Telomere Biology Disorders. *Blood*, **2021**,	2.2	2
245	Uptake and timing of bilateral and contralateral risk-reducing mastectomy in women with Li-Fraumeni syndrome. *Breast Cancer Research and Treatment*, **2021**, 1	4.4	1
244	Analysis of the Li-Fraumeni Spectrum Based on an International Germline TP53 Variant Data Set: An International Agency for Research on Cancer TP53 Database Analysis. *JAMA Oncology*, **2021**,	13.4	4
243	Gynaecological and reproductive health of women with telomere biology disorders. *British Journal of Haematology*, **2021**, 193, 1238-1246	4.5	0
242	The causes of Fanconi anemia in South Asia and the Middle East: A case series and review of the literature. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1693	2.3	0
241	RTEL1 influences the abundance and localization of TERRA RNA. *Nature Communications*, **2021**, 12, 3016	17.4	10
240	Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. *Familial Cancer*, **2021**, 1	3	
239	Alternative splicing is a developmental switch for hTERT expression. *Molecular Cell*, **2021**, 81, 2349-2360.e6	16.6	6
238	Genotype-phenotype association and variant characterization in Diamond-Blackfan anemia caused by pathogenic variants in. *Haematologica*, **2021**, 106, 1303-1310	6.6	4
237	Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. *Journal of Pediatrics*, **2021**, 230, 55-61.e4	3.6	3

236	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. *Human Mutation*, **2021**, 42, 223-236	4.7	29
235	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. *JNCI Cancer Spectrum*, **2021**, 5, pkab007	4.6	1
234	Cancer-Prone Inherited Bone Marrow Failure, Myelodysplastic, and Acute Myeloid Leukemia Syndromes **2021**, 267-314		
233	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
232	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in Patients With Osteosarcoma. *JAMA Oncology*, **2020**, 6, 724-734	13.4	60
231	XAF1 as a modifier of p53 function and cancer susceptibility. *Science Advances*, **2020**, 6, eaba3231	14.3	14
230	Population Frequency of Fanconi Pathway Gene Variants and Their Association with Survival After Hematopoietic Cell Transplantation for Severe Aplastic Anemia. *Biology of Blood and Marrow Transplantation*, **2020**, 26, 817-822	4.7	1
229	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
228	Germline mutation of , a major p53 regulator, in a familial syndrome of defective telomere maintenance. *Science Advances*, **2020**, 6, eaay3511	14.3	9
227	Disease Progression and Outcomes in Patients with Telomere Biology Disorders. *Blood*, **2020**, 136, 19-20	2.2	
226	Genetically predicted telomere length is associated with clonal somatic copy number alterations in peripheral leukocytes. *PLoS Genetics*, **2020**, 16, e1009078	6	3
225	Family Health Leaders: Lessons on Living with Li-Fraumeni Syndrome across Generations. *Family Process*, **2020**, 59, 1648-1663	3.9	6
224	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
223	Association between coffee drinking and telomere length in the Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial. *PLoS ONE*, **2020**, 15, e0226972	3.7	3
222	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
221	Pilot Study Assessing Tolerability and Metabolic Effects of Metformin in Patients With Li-Fraumeni Syndrome. *JNCI Cancer Spectrum*, **2020**, 4, pkaa063	4.6	0
220	Expansion of germline RPS20 mutation phenotype to include Diamond-Blackfan anemia. *Human Mutation*, **2020**, 41, 1918-1930	4.7	5
219	Re-equilibration of imbalanced NAD metabolism ameliorates the impact of telomere dysfunction. *EMBO Journal*, **2020**, 39, e103420	13	16

218	A pedigree-based prediction model identifies carriers of deleterious de novo mutations in families with Li-Fraumeni syndrome. *Genome Research*, **2020**, 30, 1170-1180	9.7	1
217	Pre-transplant short telomeres are associated with high mortality risk after unrelated donor haematopoietic cell transplant for severe aplastic anaemia. *British Journal of Haematology*, **2020**, 188, 309-316	4.5	5
216	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
215	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
214	Reproductive factors associated with breast cancer risk in Li-Fraumeni syndrome. *European Journal of Cancer*, **2019**, 116, 199-206	7.5	5
213	Response to: Concern regarding classification of germlineTP53 variants as likely pathogenic. *Human Mutation*, **2019**, 40, 832-833	4.7	1
212	CNS manifestations in patients with telomere biology disorders. *Neurology: Genetics*, **2019**, 5, 370	3.8	9
211	Risk of Second Primary Bone and Soft-Tissue Sarcomas Among Young Adulthood Cancer Survivors. *JNCI Cancer Spectrum*, **2019**, 3, pkz043	4.6	3
210	1q21.1 deletion and a rare functional polymorphism in siblings with thrombocytopenia-absent radius-like phenotypes. *Journal of Physical Education and Sports Management*, **2019**, 5,	2.8	1
209	Prognostic significance of pulmonary function tests in dyskeratosis congenita, a telomere biology disorder. *ERJ Open Research*, **2019**, 5,	3.5	9
208	Association between zidovudine-containing antiretroviral therapy exposure in utero and leukocyte telomere length at birth. *Aids*, **2019**, 33, 2091-2096	3.5	2
207	Understanding the evolving phenotype of vascular complications in telomere biology disorders. *Angiogenesis*, **2019**, 22, 95-102	10.6	29
206	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. *Human Mutation*, **2019**, 40, 97-105	4.7	39
205	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
204	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
203	Germline mutations in Protection of Telomeres 1 in two families with Hodgkin lymphoma. *British Journal of Haematology*, **2018**, 181, 372-377	4.5	23
202	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
201	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

200	Beginning at the ends: telomeres and human disease. *F1000Research*, **2018**, 7,	3.6	71
199	Mouse Homolog of the Human R337H Mutation Reveals Its Role in Tumorigenesis. *Cancer Research*, **2018**, 78, 5375-5383	10.1	16
198	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. *BMC Medical Genetics*, **2018**, 19, 85	2.1	10
197	Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous RTEL1 and TPH1 variants. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1432-1437	2.5	5
196	Chromosomal Aberrations and Survival after Unrelated Donor Hematopoietic Stem Cell Transplant in Patients with Fanconi Anemia. *Biology of Blood and Marrow Transplantation*, **2018**, 24, 2003-2008	4.7	7
195	Phenotypes of Diamond Blackfan Anemia Patients with RPL35A Haploinsufficiency Due to 3q29 Deletion Compared with RPL35A Single Nucleotide Variants or Small Insertion/Deletions. *Blood*, **2018**, 132, 3854-3854	2.2	3
194	Large Genomic Deletions in Shwachman-Diamond Syndrome. *Blood*, **2018**, 132, 2586-2586	2.2	0
193	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
192	Pregnancy outcomes in mothers of offspring with inherited bone marrow failure syndromes. *Pediatric Blood and Cancer*, **2018**, 65, e26757	3	4
191	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
190	Telomere Length Calibration from qPCR Measurement: Limitations of Current Method. *Cells*, **2018**, 7,	7.9	14
189	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. *Blood Advances*, **2018**, 2, 1243-1249	7.8	18
188	Myelodysplastic Syndrome, Acute Myeloid Leukemia, and Cancer Surveillance in Fanconi Anemia. *Hematology/Oncology Clinics of North America*, **2018**, 32, 657-668	3.1	26
187	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. *European Respiratory Journal*, **2017**, 49,	13.6	29
186	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
185	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
184	The genomics of inherited bone marrow failure: from mechanism to the clinic. *British Journal of Haematology*, **2017**, 177, 526-542	4.5	65
183	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

182	Estimating Mutation Carrier Probability in Families with Li-Fraumeni Syndrome Using LFSPRO. *Cancer Epidemiology Biomarkers and Prevention*, **2017**, 26, 837-844	4	7
181	Recommended Guidelines for Validation, Quality Control, and Reporting of Variants in Clinical Practice. *Cancer Research*, **2017**, 77, 1250-1260	10.1	45
180	Bone mineral density in patients with inherited bone marrow failure syndromes. *Pediatric Research*, **2017**, 82, 458-464	3.2	8
179	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
178	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
177	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. *Clinical Cancer Research*, **2017**, 23, e38-e45	12.9	245
176	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. *Clinical Cancer Research*, **2017**, 23, e23-e31	12.9	56
175	Germline and somatic genetics of osteosarcoma - connecting aetiology, biology and therapy. *Nature Reviews Endocrinology*, **2017**, 13, 480-491	15.2	225
174	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
173	Progressive reticulate skin pigmentation and anonychia in a patient with bone marrow failure. *Journal of the American Academy of Dermatology*, **2017**, 77, 1194-1198	4.5	5
172	Pediatric leukemia susceptibility disorders: manifestations and management. *Hematology American Society of Hematology Education Program*, **2017**, 2017, 242-250	3.1	7
171	Pregnancies in patients with inherited bone marrow failure syndromes in the NCI cohort. *Blood*, **2017**, 130, 1674-1676	2.2	10
170	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
169	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. *JAMA Oncology*, **2017**, 3, 1640-1645	13.4	35
168	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging: A Meta-analysis. *JAMA Oncology*, **2017**, 3, 1634-1639	13.4	107
167	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
166	Bone cancer: Is the osteosarcoma genome targetable?. *Nature Reviews Endocrinology*, **2017**, 13, 506-508	15.2	11
165	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

164	Inhibiting mitochondrial respiration prevents cancer in a mouse model of Li-Fraumeni syndrome. *Journal of Clinical Investigation*, **2017**, 127, 132-136	15.9	26
163	Effect of pre-analytic variables on the reproducibility of qPCR relative telomere length measurement. *PLoS ONE*, **2017**, 12, e0184098	3.7	30
162	Telomeres and the natural lifespan limit in humans. *Aging*, **2017**, 9, 1130-1142	5.6	53
161	Multiple Primary Cancers **2017**,		2
160	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
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	4.7	18
158	Female chromosome X mosaicism is age-related and preferentially affects the inactivated X chromosome. *Nature Communications*, **2016**, 7, 11843	17.4	59
157	Novel FANCI mutations in Fanconi anemia with VACTERL association. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 386-391	2.5	20
156	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
155	Neonatal manifestations of inherited bone marrow failure syndromes. *Seminars in Fetal and Neonatal Medicine*, **2016**, 21, 57-65	3.7	25
154	The shelterin complex and hematopoiesis. *Journal of Clinical Investigation*, **2016**, 126, 1621-9	15.9	30
153	Novel and Known Ribosomal Causes of Diamond-Blackfan Anemia Identified through Comprehensive Genomic Characterization. *Blood*, **2016**, 128, 1495-1495	2.2	1
152	Prognostic Significance of Pulmonary Function Test Abnormalities in Patients with Dyskeratosis Congenita. *Blood*, **2016**, 128, 2672-2672	2.2	2
151	Cancer in the National Cancer Institute Inherited Bone Marrow Failure Syndrome Cohort after 15 Years of Follow-up. *Blood*, **2016**, 128, 334-334	2.2	2
150	Donor Telomere Length and Outcomes after Allogeneic Unrelated Hematopoietic Cell Transplant in Patients with Acute Leukemia. *Blood*, **2016**, 128, 520-520	2.2	1
149	Germline Mutations in Patients Receiving Unrelated Donor Hematopoietic Cell Transplant for Severe Aplastic Anemia. *Blood*, **2016**, 128, 68-68	2.2	
148	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
147	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

146	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
145	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. *Pediatric Neurology*, **2016**, 56, 62-68.e1	2.9	23
144	Research participant interest in primary, secondary, and incidental genomic findings. *Genetics in Medicine*, **2016**, 18, 1218-1225	8.1	19
143	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
142	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. *Molecular Genetics & Genomic Medicine*, **2016**, 4, 475-9	2.3	16
141	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
140	Parent decision-making around the genetic testing of children for germline TP53 mutations. *Cancer* , **2015**, 121, 286-93	6.4	35
139	A Genome-Wide Scan Identifies Variants in NFIB Associated with Metastasis in Patients with Osteosarcoma. *Cancer Discovery*, **2015**, 5, 920-31	24.4	71
138	Germline TP53 variants and susceptibility to osteosarcoma. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	75
137	Bone marrow skeletal stem/progenitor cell defects in dyskeratosis congenita and telomere biology disorders. *Blood*, **2015**, 125, 793-802	2.2	26
136	Telomere length in inherited bone marrow failure syndromes. *Haematologica*, **2015**, 100, 49-54	6.6	52
135	Unraveling the pathogenesis of Hoyeraal-Hreidarsson syndrome, a complex telomere biology disorder. *British Journal of Haematology*, **2015**, 170, 457-71	4.5	79
134	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
133	Immune status of patients with inherited bone marrow failure syndromes. *American Journal of Hematology*, **2015**, 90, 702-8	7.1	20
132	Characterization of large structural genetic mosaicism in human autosomes. *American Journal of Human Genetics*, **2015**, 96, 487-97	11	77
131	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
130	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
129	Carrier screening of RTEL1 mutations in the Ashkenazi Jewish population. *Clinical Genetics*, **2015**, 88, 177-81	4	16

128	Pesticide Use and Relative Leukocyte Telomere Length in the Agricultural Health Study. *PLoS ONE*, **2015**, 10, e0133382	3.7	31
127	Toward a drug development path that targets metastatic progression in osteosarcoma. *Clinical Cancer Research*, **2014**, 20, 4200-9	12.9	103
126	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. *Nature Genetics*, **2014**, 46, 482-6	36.3	220
125	0127 Pesticide use and relative telomere length in the Agricultural Health Study. *Occupational and Environmental Medicine*, **2014**, 71, A14.3-A15	2.1	1
124	Response to androgen therapy in patients with dyskeratosis congenita. *British Journal of Haematology*, **2014**, 165, 349-57	4.5	67
123	Iodine deficiency, pollutant chemicals, and the thyroid: new information on an old problem. *Pediatrics*, **2014**, 133, 1163-6	7.4	68
122	Telomeres in molecular epidemiology studies. *Progress in Molecular Biology and Translational Science*, **2014**, 125, 113-31	4	18
121	Human telomeres and telomere biology disorders. *Progress in Molecular Biology and Translational Science*, **2014**, 125, 41-66	4	44
120	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
119	Antibody response to human papillomavirus vaccine in subjects with inherited bone marrow failure syndromes. *Vaccine*, **2014**, 32, 1169-73	4.1	12
118	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
117	Genomic clues to ethnic differences in ALL. *Blood*, **2014**, 123, 2440-2	2.2	3
116	Dubowitz syndrome is a complex comprised of multiple, genetically distinct and phenotypically overlapping disorders. *PLoS ONE*, **2014**, 9, e98686	3.7	26
115	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
114	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
113	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
112	Characterization of population-based variation and putative functional elements for the multiple-cancer susceptibility loci at 5p15.33. *F1000Research*, **2014**, 3, 231	3.6	
111	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

110	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
109	Genomic characterization of the inherited bone marrow failure syndromes. *Seminars in Hematology*, **2013**, 50, 333-47	4	57
108	Telomere length and risk of glioma. *Cancer Epidemiology*, **2013**, 37, 935-8	2.8	26
107	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
106	Common genetic variants in the 9p21 region and their associations with multiple tumours. *British Journal of Cancer*, **2013**, 108, 1378-86	8.7	40
105	Outcomes of allogeneic hematopoietic cell transplantation in patients with dyskeratosis congenita. *Biology of Blood and Marrow Transplantation*, **2013**, 19, 1238-43	4.7	86
104	Genome-wide association study identifies two susceptibility loci for osteosarcoma. *Nature Genetics*, **2013**, 45, 799-803	36.3	156
103	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
102	Erythrocyte adenosine deaminase: diagnostic value for Diamond-Blackfan anaemia. *British Journal of Haematology*, **2013**, 160, 547-54	4.5	56
101	Germline mutations of regulator of telomere elongation helicase 1, RTEL1, in Dyskeratosis congenita. *Human Genetics*, **2013**, 132, 473-80	6.3	162
100	Aplastic Anemia & MDS International Foundation (AA&MDSIF): bone marrow failure disease scientific symposium 2012. *Leukemia Research*, **2013**, 37, 848-51	2.7	2
99	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
98	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
97	Principles of Genetics and Genomics **2013**, 131-141		1
96	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. *British Journal of Haematology*, **2013**, 162, 542-6	4.5	17
95	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
94	Abstract 2532: Pediatric osteosarcoma patients are taller than average from birth to age twelve: a report from the Children Oncology Group. **2013**,		2
93	Functional Consequences Of RPS29 Germline Mutations In Diamond-Blackfan Anemia. *Blood*, **2013**, 122, 2465-2465	2.2	

92	Higher Than Expected Carrier Frequency Of The Dyskeratosis Congenita RTEL1 p.Arg1264His recessive Founder In Individuals Of Ashkenazi Jewish Ancestry. *Blood*, **2013**, 122, 1228-1228	2.2	
91	TIN2 Mutations In Dyskeratosis Congenita Cause Telomere Shortening In Induced Pluripotent Stem Cells Through Potent Dominant Negative Inhibition Of Telomerase. *Blood*, **2013**, 122, 590-590	2.2	
90	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
89	Neuropsychiatric conditions among patients with dyskeratosis congenita: a link with telomere biology?. *Psychosomatics*, **2012**, 53, 230-5	2.6	19
88	Hereditary and environmental epidemiology of sarcomas. *Clinical Sarcoma Research*, **2012**, 2, 13	2.5	2
87	Detectable clonal mosaicism and its relationship to aging and cancer. *Nature Genetics*, **2012**, 44, 651-8	36.3	409
86	The relationship between DNA methylation and telomere length in dyskeratosis congenita. *Aging Cell*, **2012**, 11, 24-8	9.9	23
85	Worldwide genetic structure in 37 genes important in telomere biology. *Heredity*, **2012**, 108, 124-33	3.6	10
84	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
83	Connecting complex disorders through biology. *Nature Genetics*, **2012**, 44, 238-40	36.3	20
82	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
81	Telomere length is associated with disease severity and declines with age in dyskeratosis congenita. *Haematologica*, **2012**, 97, 353-9	6.6	145
80	Response to Androgen Therapy and Side Effects in Patients with Dyskeratosis Congenita.. *Blood*, **2012**, 120, 2361-2361	2.2	1
79	On the interplay of telomeres, nevi and the risk of melanoma. *PLoS ONE*, **2012**, 7, e52466	3.7	15
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