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	Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. Familial Cancer, 2022, 21, 333-336.	0.9	1
2	Uptake and timing of bilateral and contralateral risk-reducing mastectomy in women with Liâ€“Fraumeni syndrome. Breast Cancer Research and Treatment, 2022, 191, 159-167.	1.1	5
3	Disease progression and clinical outcomes in telomere biology disorders. Blood, 2022, 139, 1807-1819.	0.6	34
4	Lipoprotein particle alterations due to androgen therapy in individuals with dyskeratosis congenita. EBioMedicine, 2022, 75, 103760.	2.7	1
5	Spectrum and Incidence of Skin Cancer among Individuals with Li-Fraumeni Syndrome. Journal of Investigative Dermatology, 2022, 142, 2534-2537.e1.	0.3	1
6	Telomere biology disorders gain a family member. Blood, 2022, 139, 957-959.	0.6	1
7	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
8	Shwachman Diamond syndrome: narrow genotypic spectrum and variable clinical features. Pediatric Research, 2022, , .	1.1	5
9	Fundamental immuneâ€“oncogenicity trade-offs define driver mutationÂfitness. Nature, 2022, 606, 172-179.	13.7	23
10	Telomere length and epigenetic clocks as markers of cellular aging: a comparative study. GeroScience, 2022, 44, 1861-1869.	2.1	18
11	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
12	Genetic testing in severe aplastic anemia is required forÂoptimal hematopoietic cell transplant outcomes. Blood, 2022, 140, 909-921.	0.6	18
13	Genotype-phenotype association and variant characterization in Diamond-Blackfan anemia caused by pathogenic variants in <i>RPL35A</i>. Haematologica, 2021, 106, 1303-1310.	1.7	12
14	Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. Journal of Pediatrics, 2021, 230, 55-61.e4.	0.9	14
15	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	1.1	81
16	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in the Childhood Cancer Survivor Study. JNCI Cancer Spectrum, 2021, 5, pkab007.	1.4	11
17	Cancer-Prone Inherited Bone Marrow Failure, Myelodysplastic, and Acute Myeloid Leukemia Syndromes. , 2021, , 267-314.		0
18	Gynaecological and reproductive health of women with telomere biology disorders. British Journal of Haematology, 2021, 193, 1238-1246.	1.2	5

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19	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.	0.6	2
20	RTEL1 influences the abundance and localization of TERRA RNA. Nature Communications, 2021, 12, 3016.	5.8	30
21	Alternative splicing is a developmental switch for hTERT expression. Molecular Cell, 2021, 81, 2349-2360.e6.	4.5	19
22	DNA-methylation-based telomere length estimator: comparisons with measurements from flow FISH and qPCR. Aging, 2021, 13, 14675-14686.	1.4	11
23	Pathogenic germline IKZF1 variant alters hematopoietic gene expression profiles. Journal of Physical Education and Sports Management, 2021, 7, a006015.	0.5	5
24	Analysis of the Li-Fraumeni Spectrum Based on an International Germline TP53 Variant Data Set. JAMA Oncology, 2021, 7, 1800.	3.4	55
25	Whole Exome Sequencing in Severe Aplastic Anemia Identifies Unrecognized Inherited Subset with Inferior Survival after Hematopoietic Cell Transplant. Blood, 2021, 138, 605-605.	0.6	0
26	Genomic-Based Machine Learning Towards Prediction of the Etiology of Bone Marrow Failure Syndromes. Blood, 2021, 138, 2182-2182.	0.6	1
27	Germline-Somatic Interactions in Myelofibrosis Susceptibility. Blood, 2021, 138, 313-313.	0.6	0
28	Quantification of Discordant Variant Interpretations in a Large Family-Based Study of Li-Fraumeni Syndrome. JCO Precision Oncology, 2021, 5, 1727-1737.	1.5	3
29	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
30	Preâ€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.	1.2	9
31	Family Health Leaders: Lessons on Living with Liâ€Fraumeni Syndrome across Generations. Family Process, 2020, 59, 1648-1663.	1.4	10
32	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
33	Association between coffee drinking and telomere length in the Prostate, Lung, Colorectal, and Ovarian Cancer Screening Trial. PLoS ONE, 2020, 15, e0226972.	1.1	5
34	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
35	Pilot Study Assessing Tolerability and Metabolic Effects of Metformin in Patients With Li-Fraumeni Syndrome. JNCI Cancer Spectrum, 2020, 4, pkaa063.	1.4	6
36	Expansion of germline RPS20 mutation phenotype to include Diamondâ€“Blackfan anemia. Human Mutation, 2020, 41, 1918-1930.	1.1	13

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37	Reâ€equilibration of imbalanced NAD metabolism ameliorates the impact of telomere dysfunction. EMBO Journal, 2020, 39, e103420.	3.5	42
38	A pedigree-based prediction model identifies carriers of deleterious de novo mutations in families with Li-Fraumeni syndrome. Genome Research, 2020, 30, 1170-1180.	2.4	4
39	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
40	Frequency of Pathogenic Germline Variants in Cancer-Susceptibility Genes in Patients With Osteosarcoma. JAMA Oncology, 2020, 6, 724.	3.4	139
41	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	4.7	37
42	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.	2.0	6
43	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
44	Germline mutation of MDM4 , a major p53 regulator, in a familial syndrome of defective telomere maintenance. Science Advances, 2020, 6, eaay3511.	4.7	25
45	Genetically predicted telomere length is associated with clonal somatic copy number alterations in peripheral leukocytes. PLoS Genetics, 2020, 16, e1009078.	1.5	14
46	Disease Progression and Outcomes in Patients with Telomere Biology Disorders. Blood, 2020, 136, 19-20.	0.6	0
47	CNS manifestations in patients with telomere biology disorders. Neurology: Genetics, 2019, 5, 370.	0.9	17
48	Risk of Second Primary Bone and Softâ€“Tissue Sarcomas Among Young Adulthood Cancer Survivors. JNCI Cancer Spectrum, 2019, 3, pkz043.	1.4	7
49	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
50	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
51	Reproductive factors associated with breast cancer risk in Liâ€“Fraumeni syndrome. European Journal of Cancer, 2019, 116, 199-206.	1.3	10
52	Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. Human Mutation, 2019, 40, 832-833.	1.1	1
53	1q21.1 deletion and a rare functional polymorphism in siblings with thrombocytopenia-absent radiusâ€“like phenotypes. Journal of Physical Education and Sports Management, 2019, 5, a004564.	0.5	4
54	Prognostic significance of pulmonary function tests in dyskeratosis congenita, a telomere biology disorder. ERJ Open Research, 2019, 5, 00209-2019.	1.1	13

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55	Association between zidovudine-containing antiretroviral therapy exposure in utero and leukocyte telomere length at birth. Aids, 2019, 33, 2091-2096.	1.0	2
56	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	3.7	45
57	Variable population prevalence estimates of germline TP53 variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105.	1.1	66
58	Donor telomere length and causes of death after unrelated hematopoietic cell transplantation in patients with marrow failure. Blood, 2018, 131, 2393-2398.	0.6	15
59	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.	1.3	13
60	Germline mutations in Protection of Telomeres 1 in two families with Hodgkin lymphoma. British Journal of Haematology, 2018, 181, 372-377.	1.2	48
61	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
62	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
63	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
64	Pregnancy outcomes in mothers of offspring with inherited bone marrow failure syndromes. Pediatric Blood and Cancer, 2018, 65, e26757.	0.8	11
65	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.	3.6	15
66	Telomere Length Calibration from qPCR Measurement: Limitations of Current Method. Cells, 2018, 7, 183.	1.8	23
67	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. Blood Advances, 2018, 2, 1243-1249.	2.5	30
68	Myelodysplastic Syndrome, Acute Myeloid Leukemia, and Cancer Surveillance in Fanconi Anemia. Hematology/Oncology Clinics of North America, 2018, 32, 657-668.	0.9	42
69	Beginning at the ends: telomeres and human disease. F1000Research, 2018, 7, 524.	0.8	105
70	Mouse Homolog of the Human TP53 R337H Mutation Reveals Its Role in Tumorigenesis. Cancer Research, 2018, 78, 5375-5383.	0.4	24
71	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. BMC Medical Genetics, 2018, 19, 85.	2.1	16
72	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.	0.7	7

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73	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.	2.0	9
74	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.	0.6	3
75	Large Genomic Deletions in Shwachman-Diamond Syndrome. Blood, 2018, 132, 2586-2586.	0.6	2
76	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. European Respiratory Journal, 2017, 49, 1601640.	3.1	41
77	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
78	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
79	The genomics of inherited bone marrow failure: from mechanism to the clinic. British Journal of Haematology, 2017, 177, 526-542.	1.2	89
80	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
81	Estimating TP53 Mutation Carrier Probability in Families with Liâ€“Fraumeni Syndrome Using LFSPRO. Cancer Epidemiology Biomarkers and Prevention, 2017, 26, 837-844.	1.1	14
82	Recommended Guidelines for Validation, Quality Control, and Reporting of TP53 Variants in Clinical Practice. Cancer Research, 2017, 77, 1250-1260.	0.4	68
83	Bone mineral density in patients with inherited bone marrow failure syndromes. Pediatric Research, 2017, 82, 458-464.	1.1	10
84	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.	2.0	9
85	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
86	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45.	3.2	358
87	Recommendations for Childhood Cancer Screening and Surveillance in DNA Repair Disorders. Clinical Cancer Research, 2017, 23, e23-e31.	3.2	93
88	Germline and somatic genetics of osteosarcoma â€" connecting aetiology, biology and therapy. Nature Reviews Endocrinology, 2017, 13, 480-491.	4.3	319
89	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
90	Progressive reticulate skin pigmentation and anonychia in a patient with bone marrow failure. Journal of the American Academy of Dermatology, 2017, 77, 1194-1198.	0.6	8

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91	Pregnancies in patients with inherited bone marrow failure syndromes in the NCI cohort. Blood, 2017, 130, 1674-1676.	0.6	15
92	Higher-than-expected population prevalence of potentially pathogenic germlineTP53variants in individuals unselected for cancer history. Human Mutation, 2017, 38, 1723-1730.	1.1	40
93	Prevalence of Cancer at Baseline Screening in the National Cancer Institute Li-Fraumeni Syndrome Cohort. JAMA Oncology, 2017, 3, 1640.	3.4	43
94	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634.	3.4	148
95	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
96	Is the osteosarcoma genome targetable?. Nature Reviews Endocrinology, 2017, 13, 506-508.	4.3	16
97	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
98	Pediatric leukemia susceptibility disorders: manifestations and management. Hematology American Society of Hematology Education Program, 2017, 2017, 242-250.	0.9	11
99	Multiple Primary Cancers. , 2017, , .		3
100	Effect of pre-analytic variables on the reproducibility of qPCR relative telomere length measurement. PLoS ONE, 2017, 12, e0184098.	1.1	55
101	Telomeres and the natural lifespan limit in humans. Aging, 2017, 9, 1130-1142.	1.4	82
102	Bone Cancers. , 2017, , .		0
103	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
104	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
105	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
106	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. Pediatric Neurology, 2016, 56, 62-68.e1.	1.0	29
107	Research participant interest in primary, secondary, and incidental genomic findings. Genetics in Medicine, 2016, 18, 1218-1225.	1.1	24
108	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

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109	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. Molecular Genetics & Genomic Medicine, 2016, 4, 475-479.	0.6	20
110	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
111	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
112	Female chromosome X mosaicism is age-related and preferentially affects the inactivated X chromosome. Nature Communications, 2016, 7, 11843.	5.8	86
113	Novel FANCI mutations in Fanconi anemia with VACTERL association. American Journal of Medical Genetics, Part A, 2016, 170, 386-391.	0.7	25
114	Easing the Burden: Describing the Role of Social, Emotional and Spiritual Support in Research Families with Lia€Fraumeni Syndrome. Journal of Genetic Counseling, 2016, 25, 529-542.	0.9	24
115	Neonatal manifestations of inherited bone marrow failure syndromes. Seminars in Fetal and Neonatal Medicine, 2016, 21, 57-65.	1.1	37
116	The shelterin complex and hematopoiesis. Journal of Clinical Investigation, 2016, 126, 1621-1629.	3.9	40
117	Inhibiting mitochondrial respiration prevents cancer in a mouse model of Li-Fraumeni syndrome. Journal of Clinical Investigation, 2016, 127, 132-136.	3.9	39
118	Novel and Known Ribosomal Causes of Diamond-Blackfan Anemia Identified through Comprehensive Genomic Characterization. Blood, 2016, 128, 1495-1495.	0.6	1
119	Prognostic Significance of Pulmonary Function Test Abnormalities in Patients with Dyskeratosis Congenita. Blood, 2016, 128, 2672-2672.	0.6	2
120	Cancer in the National Cancer Institute Inherited Bone Marrow Failure Syndrome Cohort after 15 Years of Follow-up. Blood, 2016, 128, 334-334.	0.6	2
121	Donor Telomere Length and Outcomes after Allogeneic Unrelated Hematopoietic Cell Transplant in Patients with Acute Leukemia. Blood, 2016, 128, 520-520.	0.6	1
122	Germline Mutations in Patients Receiving Unrelated Donor Hematopoietic Cell Transplant for Severe Aplastic Anemia. Blood, 2016, 128, 68-68.	0.6	0
123	Bone marrow skeletal stem/progenitor cell defects in dyskeratosis congenita and telomere biology disorders. Blood, 2015, 125, 793-802.	0.6	31
124	Telomere length in inherited bone marrow failure syndromes. Haematologica, 2015, 100, 49-54.	1.7	63
125	Unraveling the pathogenesis of Hoyeraala€"Hreidarsson syndrome, a complex telomere biology disorder. British Journal of Haematology, 2015, 170, 457-471.	1.2	105
126	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

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127	Immune status of patients with inherited bone marrow failure syndromes. American Journal of Hematology, 2015, 90, 702-708.	2.0	34
128	Characterization of Large Structural Genetic Mosaicism in Human Autosomes. American Journal of Human Genetics, 2015, 96, 487-497.	2.6	101
129	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
130	Reduced Serum Levels of Anti-MÃ¼llerian Hormone in Females With Inherited Bone Marrow Failure Syndromes. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E197-E203.	1.8	14
131	Carrier screening of RTEL1 mutations in the Ashkenazi Jewish population. Clinical Genetics, 2015, 88, 177-181.	1.0	18
132	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
133	Parent decisionâ€making around the genetic testing of children for germline TP53 mutations. Cancer, 2015, 121, 286-293.	2.0	41
134	A Genome-Wide Scan Identifies Variants in NFIB Associated with Metastasis in Patients with Osteosarcoma. Cancer Discovery, 2015, 5, 920-931.	7.7	88
135	Germline TP53 Variants and Susceptibility to Osteosarcoma. Journal of the National Cancer Institute, 2015, 107, .	3.0	109
136	Pesticide Use and Relative Leukocyte Telomere Length in the Agricultural Health Study. PLoS ONE, 2015, 10, e0133382.	1.1	42
137	Abstract 2749: Cumulative cancer risk in the NCI Li-Fraumeni Syndrome Cohort. , 2015, , .		0
138	Dubowitz Syndrome Is a Complex Comprised of Multiple, Genetically Distinct and Phenotypically Overlapping Disorders. PLoS ONE, 2014, 9, e98686.	1.1	29
139	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
140	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
141	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
142	Toward a Drug Development Path That Targets Metastatic Progression in Osteosarcoma. Clinical Cancer Research, 2014, 20, 4200-4209.	3.2	127
143	Rare missense variants in POT1 predispose to familial cutaneous malignant melanoma. Nature Genetics, 2014, 46, 482-486.	9.4	283
144	0127â€…Pesticide use and relative telomere length in the Agricultural Health Study. Occupational and Environmental Medicine, 2014, 71, A14.3-A15.	1.3	1

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145	Response to androgen therapy in patients with dyskeratosis congenita. British Journal of Haematology, 2014, 165, 349-357.	1.2	89
146	Iodine Deficiency, Pollutant Chemicals, and the Thyroid: New Information on an Old Problem. Pediatrics, 2014, 133, 1163-1166.	1.0	82
147	Telomeres in Molecular Epidemiology Studies. Progress in Molecular Biology and Translational Science, 2014, 125, 113-131.	0.9	23
148	Human Telomeres and Telomere Biology Disorders. Progress in Molecular Biology and Translational Science, 2014, 125, 41-66.	0.9	60
149	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
150	Donor Telomere Length Predicts Recipient Survival after Allogeneic Hematopoietic Cell Transplantation in Patients with Bone Marrow Failure Syndromes. Biology of Blood and Marrow Transplantation, 2014, 20, S33-S34.	2.0	0
151	Antibody response to human papillomavirus vaccine in subjects with inherited bone marrow failure syndromes. Vaccine, 2014, 32, 1169-1173.	1.7	13
152	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
153	Genomic clues to ethnic differences in ALL. Blood, 2014, 123, 2440-2442.	0.6	3
154	Characterization of population-based variation and putative functional elements for the multiple-cancer susceptibility loci at 5p15.33. F1000Research, 2014, 3, 231.	0.8	0
155	Abstract 29: Design of a phase I chemoprevention study of metformin and Li-Fraumeni syndrome (LFS). , 2014, , .		1
156	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
157	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
158	Genomic Characterization of the Inherited Bone Marrow Failure Syndromes. Seminars in Hematology, 2013, 50, 333-347.	1.8	69
159	Telomere length and risk of glioma. Cancer Epidemiology, 2013, 37, 935-938.	0.8	28
160	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
161	The Long and Short of Telomeres and Cancer Association Studies. Journal of the National Cancer Institute, 2013, 105, 448-449.	3.0	25
162	Common genetic variants in the 9p21 region and their associations with multiple tumours. British Journal of Cancer, 2013, 108, 1378-1386.	2.9	55

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163	Outcomes of Allogeneic Hematopoietic Cell Transplantation in Patients with Dyskeratosis Congenita. Biology of Blood and Marrow Transplantation, 2013, 19, 1238-1243.	2.0	108
164	Genome-wide association study identifies two susceptibility loci for osteosarcoma. Nature Genetics, 2013, 45, 799-803.	9.4	181
165	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
166	Erythrocyte adenosine deaminase: diagnostic value for Diamondâ€Blackfan anaemia. British Journal of Haematology, 2013, 160, 547-554.	1.2	76
167	Germline mutations of regulator of telomere elongation helicase 1, RTEL1, in Dyskeratosis congenita. Human Genetics, 2013, 132, 473-480.	1.8	198
168	Aplastic Anemia & MDS International Foundation (AA&MDSIF): Bone marrow failure disease scientific symposium 2012. Leukemia Research, 2013, 37, 848-851.	0.4	2
169	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
170	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
171	Principles of Genetics and Genomics. , 2013, , 131-141.		1
172	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. British Journal of Haematology, 2013, 162, 542-546.	1.2	21
173	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
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