Alison A Bertuch

List of Publications by Year
in descending order

101 papers	3,497 citations	136950 32 h-index	155660 55 g-index
106 all docs	106 docs citations	106 times ranked	5758 citing authors

#	Article	IF	Citations
1	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
2	The genetics and clinical manifestations of telomere biology disorders. Genetics in Medicine, 2010, 12, 753-764.	2.4	204
3	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. Journal of the National Cancer Institute, 2015, 107, 384.	6.3	172
4	The molecular genetics of the telomere biology disorders. RNA Biology, 2016, 13, 696-706.	3.1	144
5	Dyskeratosis congenita as a disorder of telomere maintenance. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2012, 730, 43-51.	1.0	116
6	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. Blood Advances, 2019, 3, 2962-2979.	5.2	110
7	Distinct genetic pathways define pre-malignant versus compensatory clonal hematopoiesis in Shwachman-Diamond syndrome. Nature Communications, 2021, 12, 1334.	12.8	103
8	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. Human Molecular Genetics, 2005, 14, 3723-3740.	2.9	94
9	A role for heterochromatin protein 1Î³ at human telomeres. Genes and Development, 2011, 25, 1807-1819.	5.9	93
10	The Ku Heterodimer Performs Separable Activities at Double-Strand Breaks and Chromosome Termini. Molecular and Cellular Biology, 2003, 23, 8202-8215.	2.3	91
11	Somatic mutations and clonal hematopoiesis in congenital neutropenia. Blood, 2018, 131, 408-416.	1.4	91
12	Distinct faces of the Ku heterodimer mediate DNA repair and telomeric functions. Nature Structural and Molecular Biology, 2007, 14, 301-307.	8.2	88
13	Proposal for the clinical detection and management of patients and their family members with familial myelodysplastic syndrome/acute leukemia predisposition syndromes. Leukemia and Lymphoma, 2013, 54, 28-35.	1.3	88
14	EXO1 Contributes to Telomere Maintenance in Both Telomerase-Proficient and Telomerase-Deficient Saccharomyces cerevisiae. Genetics, 2004, 166, 1651-1659.	2.9	80
15	Compound Heterozygous CORO1A Mutations in Siblings with a Mucocutaneous-Immunodeficiency Syndrome of Epidermodysplasia Verruciformis-HPV, Molluscum Contagiosum and Granulomatous Tuberculoid Leprosy. Journal of Clinical Immunology, 2014, 34, 871-890.	3.8	78
16	Poly(ADP-ribose) polymerase inhibitor ABT-888 potentiates the cytotoxic activity of temozolomide in leukemia cells: influence of mismatch repair status and O6-methylguanine-DNA methyltransferase activity. Molecular Cancer Therapeutics, 2009, 8, 2232-2242.	4.1	77
17	Structural Insights into Yeast Telomerase Recruitment to Telomeres. Cell, 2018, 172, 331-343.e13.	28.9	76
18	Three novel truncating TINF2 mutations causing severe dyskeratosis congenita in early childhood. Clinical Genetics, 2012, 81, 470-478.	2.0	74

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19	Evaluation of Patients and Families With Concern for Predispositions to Hematologic Malignancies Within the Hereditary Hematologic Malignancy Clinic (HHMC). Clinical Lymphoma, Myeloma and Leukemia, 2016, 16, 417-428.e2.	0.4	74
20	Short telomeres: from dyskeratosis congenita to sporadic aplastic anemia and malignancy. Translational Research, 2013, 162, 353-363.	5.0	73
21	Consider the workhorse: Nonhomologous end-joining in budding yeast. Biochemistry and Cell Biology, 2016, 94, 396-406.	2.0	56
22	The maintenance and masking of chromosome termini. Current Opinion in Cell Biology, 2006, 18, 247-253.	5.4	52
23	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
24	TRF2 Interaction with Ku Heterotetramerization Interface Gives Insight into c-NHEJ Prevention at Human Telomeres. Cell Reports, 2013, 5, 194-206.	6.4	51
25	Segregating YKU80 and TLC1 Alleles Underlying Natural Variation in Telomere Properties in Wild Yeast. PLoS Genetics, 2009, 5, e1000659.	3.5	46
26	Understanding the evolving phenotype of vascular complications in telomere biology disorders. Angiogenesis, 2019, 22, 95-102.	7.2	45
27	Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study. Haematologica, 2019, 104, 1974-1983.	3.5	43
28	Clinical, histopathologic, and genetic features of pediatric primary myelofibrosisâ€"An entity different from adults. American Journal of Hematology, 2012, 87, 461-464.	4.1	42
29	Telomere dysfunction activates YAP1 to drive tissue inflammation. Nature Communications, 2020, 11, 4766.	12.8	42
30	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. European Respiratory Journal, 2017, 49, 1601640.	6.7	41
31	Telomeres and double-strand breaks: trying to make ends meet. Trends in Cell Biology, 1998, 8, 339-342.	7.9	40
32	Infantile fibrosarcoma: Clinical and histologic responses to cytotoxic chemotherapy. Pediatric Blood and Cancer, 2009, 53, 23-27.	1.5	38
33	Ku Must Load Directly onto the Chromosome End in Order to Mediate Its Telomeric Functions. PLoS Genetics, 2011, 7, e1002233.	3.5	35
34	Features of Hepatitis in Hepatitisâ€associated Aplastic Anemia. Journal of Pediatric Gastroenterology and Nutrition, 2017, 64, e7-e12.	1.8	32
35	Which end: dissecting Ku's function at telomeres and double-strand breaks. Genes and Development, 2003, 17, 2347-2350.	5.9	31
36	Diagnosis and treatment of pediatric acquired aplastic anemia (AAA): An initial survey of the North American Pediatric Aplastic Anemia Consortium (NAPAAC). Pediatric Blood and Cancer, 2014, 61, 869-874.	1.5	31

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37	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. Blood Advances, 2018, 2, 1243-1249.	5.2	30
38	EXO1 Contributes to Telomere Maintenance in Both Telomerase-Proficient and Telomerase-Deficient Saccharomyces cerevisiae. Genetics, 2004, 166, 1651-1659.	2.9	29
39	The Association of yKu With Subtelomeric Core X Sequences Prevents Recombination Involving Telomeric Sequences. Genetics, 2009, 183, 453-467.	2.9	28
40	A homozygous telomerase T-motif variant resulting in markedly reduced repeat addition processivity in siblings with Hoyeraal Hreidarsson syndrome. Blood, 2013, 121, 3586-3593.	1.4	28
41	Regulation of Ku-DNA Association by Yku70 C-terminal Tail and SUMO Modification. Journal of Biological Chemistry, 2014, 289, 10308-10317.	3.4	28
42	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
43	The C-Terminal Extension Unique to the Long Isoform of the Shelterin Component TIN2 Enhances Its Interaction with TRF2 in a Phosphorylation- and Dyskeratosis Congenita Cluster-Dependent Fashion. Molecular and Cellular Biology, 2018, 38, .	2.3	25
44	A Single Institution Experience With Pediatric Nasopharyngeal Carcinoma: High Incidence of Toxicity Associated With Platinum-based Chemotherapy Plus IMRT. Journal of Pediatric Hematology/Oncology, 2007, 29, 500-505.	0.6	23
45	Somatic mutations in children with GATA2-associated myelodysplastic syndrome who lack other features of GATA2 deficiency. Blood Advances, 2017, 1, 443-448.	5.2	23
46	PSTPIP1â€associated myeloidâ€related proteinemia inflammatory syndrome: A rare cause of childhood neutropenia associated with systemic inflammation and hyperzincemia. Pediatric Blood and Cancer, 2019, 66, e27439.	1.5	23
47	Ku's essential role in keeping telomeres intact. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 12217-12218.	7.1	19
48	An S/T-Q cluster domain census unveils new putative targets under Tel1/Mec1 control. BMC Genomics, 2012, 13, 664.	2.8	19
49	Shorter Remission Telomere Length Predicts Delayed Neutrophil Recovery After Acute Myeloid Leukemia Therapy: A Report From the Childrenâ€™s Oncology Group. Journal of Clinical Oncology, 2016, 34, 3766-3772.	1.6	17
50	The Principal Role of Ku in Telomere Length Maintenance Is Promotion of Est1 Association with Telomeres. Genetics, 2014, 197, 1123-1136.	2.9	16
51	Changes Mimicking New Leptomeningeal Disease After Intensity-Modulated Radiotherapy for Medulloblastoma. International Journal of Radiation Oncology Biology Physics, 2009, 73, 214-221.	0.8	15
52	Hematologic presentation and the role of untargeted metabolomics analysis in monitoring treatment for riboflavin transporter deficiency. American Journal of Medical Genetics, Part A, 2020, 182, 2781-2787.	1.2	15
53	Hematopoiesis under telomere attrition at the single-cell resolution. Nature Communications, 2021, 12, 6850.	12.8	15
54	From incomplete penetrance with normal telomere length to severe disease and telomere shortening in a family with monoallelic and biallelic PARN pathogenic variants. Human Mutation, 2019, 40, 2414-2429.	2.5	14

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55	Diagnostic workâ€up for severe aplastic anemia in children: Consensus of the <scp>North American Pediatric Aplastic Anemia Consortium</scp>. American Journal of Hematology, 2021, 96, 1491-1504.	4.1	14
56	Expansion of germline RPS20 mutation phenotype to include Diamondâ€“Blackfan anemia. Human Mutation, 2020, 41, 1918-1930.	2.5	13
57	Ku DNA End-Binding Activity Promotes Repair Fidelity and Influences End-Processing During Nonhomologous End-Joining in Saccharomyces cerevisiae. Genetics, 2018, 209, 115-128.	2.9	12
58	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. Blood, 2021, 138, 2441-2445.	1.4	12
59	Modeling growth and telomere dynamics in Saccharomyces cerevisiae. Journal of Theoretical Biology, 2010, 263, 353-359.	1.7	11
60	A study assessing the feasibility of randomization of pediatric and young adult patients between matched unrelated donor bone marrow transplantation and immuneâ€suppressive therapy for newly diagnosed severe aplastic anemia: A joint pilot trial of the North American Pediatric Aplastic Anemia Consortium and the Pediatric Transplantation and Cellular Therapy Consortium. Pediatric Blood and Cancer, 2020, 67, e28444.	1.5	11
61	Thrombopoietin Measurement as a Key Component in the Evaluation of Pediatric Thrombocytosis. Pediatric Blood and Cancer, 2016, 63, 1484-1487.	1.5	10
62	Telomeres: The Molecular Events Driving End-To-End Fusions. Current Biology, 2002, 12, R738-R740.	3.9	9
63	Structural variation and missense mutation in SBDSassociated with Shwachman-Diamond syndrome. BMC Medical Genetics, 2014, 15, 64.	2.1	9
64	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. Blood, 2018, 132, 658-662.	1.4	9
65	Adenosine Deaminase 2 Deficiency As a Cause of Pure Red Cell Aplasia Mimicking Diamond Blackfan Anemia. Blood, 2015, 126, 3615-3615.	1.4	9
66	Measuring relative telomere length: Is tissue an issue?. Aging, 2010, 2, 756-757.	3.1	9
67	Identification and characterization of novel ACD variants: modulation of TPP1 protein level offsets the impact of germline loss-of-function variants on telomere length. Journal of Physical Education and Sports Management, 2021, 7, a005454.	1.2	8
68	Expansion of the clinical phenotype of <scp>GALE</scp> deficiency. American Journal of Medical Genetics, Part A, 2021, 185, 3118-3121.	1.2	8
69	Standardized highâ€sensitivity flow cytometry testing for paroxysmal nocturnal hemoglobinuria in children with acquired bone marrow failure disorders: A single center US study. Cytometry Part B - Clinical Cytometry, 2018, 94, 699-704.	1.5	7
70	The Way to the End Matters: The Role of Telomerase in Tumor Progression. Cell Cycle, 2003, 2, 36-38.	2.6	6
71	Editing TINF2 as a potential therapeutic approach to restore telomere length in dyskeratosis congenita. Blood, 2022, 140, 608-618.	1.4	5
72	Abdominal undifferentiated small round cell tumor with unique translocation (X;19)(q13;q13.3). Pediatric Blood and Cancer, 2010, 54, 1041-1044.	1.5	4

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73	Tryptophan-Dependent Control of Colony Formation After DNA Damage via Sea3-Regulated TORC1 Signaling in Saccharomyces cerevisiae. G3: Genes, Genomes, Genetics, 2015, 5, 1379-1389.	1.8	4
74	Dyskeratosis Congenita and the Telomere Biology Disorders. Pediatric Oncology, 2018, , 111-135.	0.5	4
75	Loss of Kuâ€™s DNA end binding activity affects telomere length via destabilizing telomere-bound Est1 rather than altering TLC1 homeostasis. Scientific Reports, 2019, 9, 10607.	3.3	4
76	Clinical and functional characterization of telomerase variants in patients with pediatric acute myeloid leukemia/myelodysplastic syndrome. Leukemia, 2021, 35, 269-273.	7.2	4
77	Severe therapyâ€related toxicities after treatment for Hodgkin lymphoma due to a pathogenic TERT variant and shortened telomeres. Pediatric Blood and Cancer, 2019, 66, e27779.	1.5	3
78	Monozygotic twins with non-Down syndrome associated MLL-rearranged hematologic malignancy and megakaryoblastic differentiation. Leukemia and Lymphoma, 2019, 60, 1083-1086.	1.3	3
79	The germline p53 activation syndrome: A new patient further refines the clinical phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 2204-2208.	1.2	3
80	Matched Unrelated Allogeneic Stem Cell Transplantation for Congenital Amegakaryocytic Thrombocytopenia: Texas Children's Hospital Experience. Biology of Blood and Marrow Transplantation, 2016, 22, S237.	2.0	1
81	De Novo RPS20 Mutations in Diamond Blackfan Anemia. Blood, 2014, 124, 2667-2667.	1.4	1
82	A Targeted Next-Generation Sequencing Mutation Panel for Pediatric Acute Myeloid Leukemia and Myelodysplastic Syndrome (MDS) Detects Potential Additional Driver Mutations in Pediatric GATA2-MDS. Blood, 2015, 126, 1679-1679.	1.4	1
83	Primary Myelofibrosis In Children. Blood, 2010, 116, 3079-3079.	1.4	1
84	GATA2 Mutations In Nonsyndromic Pediatric Myelodysplastic Syndrome. Blood, 2013, 122, 2778-2778.	1.4	1
85	Outcome after Stem Cell Transplant in Patients with Dyskeratosis Congenita. Biology of Blood and Marrow Transplantation, 2014, 20, S178-S179.	2.0	0
86	A new mutant at the end: TPP1, telomeres, and BMF. Blood, 2014, 124, 2757-2758.	1.4	0
87	High-Sensitivity Flow Cytometry Testing for Paroxysmal Nocturnal Hemoglobinuria in Children with Cytopenia: A Single Center Study. Blood, 2011, 118, 2398-2398.	1.4	0
88	Clinical and Molecular Characterization of Germline Telomerase Reverse Transcriptase (TERT) Variants in Children with Acute Myeloid Leukemia (AML),. Blood, 2011, 118, 3571-3571.	1.4	0
89	Constitutional Telomerase-Associated Gene Variants Associated with Chemotherapy-Related Toxicities in Pediatric Acute Myeloid Leukemia (AML). Blood, 2012, 120, 1403-1403.	1.4	0
90	A Homozygous Telomerase Reverse Transcriptase T Motif Variant Resulting in Markedly Reduced Telomerase Repeat Addition Processivity in Siblings with Hoyeraal Hreidarsson Syndrome. Blood, 2012, 120, 1272-1272.	1.4	0

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91	Constitutional Telomerase-Associated Gene Variants in Pediatric Acute Myeloid Leukemia (AML). Blood, 2012, 120, 1408-1408.	1.4	0
92	Constitutional Telomerase-Associated Gene Variants In Pediatric Acute Myeloid Leukemia (AML) and In Association With Chemotherapy-Related Toxicities. Blood, 2013, 122, 1310-1310.	1.4	0
93	Aplastic Anemia and Advocacy for Some Uniquely Pediatric Issues. , 2014, 11, .		0
94	Matched Unrelated Allogeneic Stem Cell Transplantation for Patients with Congenital Amegakaryocytic Thrombocytopenia: Texas Children`s Hospital Experience. Blood, 2015, 126, 5529-5529.	1.4	0
95	A Novel Radiosensitivity Phenotype in Shwachman-Diamond Syndrome Is Mediated By ER Stress Response. Blood, 2015, 126, 3618-3618.	1.4	0
96	Myeloid Malignancy Variant Curation Expert Panel: An ASH-Sponsored Clingen Expert Panel to Optimize and Validate Acmg/AMP Variant Interpretation Guidelines for Genes Associated with Inherited Myeloid Neoplasms. Blood, 2018, 132, 5849-5849.	1.4	0
97	The Shwachman-Diamond Syndrome Registry: Hematologic Complications. Blood, 2018, 132, 3871-3871.	1.4	0
98	Cells Deficient in the Shwachman-Diamond Syndrome Protein SBDS or the Diamond-Blackfan Anemia Protein RPS19 Have Impaired Homologous Recombination. Blood, 2019, 134, 104-104.	1.4	0
99	Genetic Dissection of Dyskeratosis Congenita-Causing Mutations in TINF2 Using Human Pluripotent and Hematopoietic Stem Cell Models. Blood, 2021, 138, 2178-2178.	1.4	0
100	Distinct Genetic Pathways Define Leukemia Predisposition Versus Adaptive Clonal Hematopoiesis in Shwachman-Diamond Syndrome. Blood, 2020, 136, 35-36.	1.4	0
101	Towards Identifying the Target of Autoimmunity in Aplastic Anemia. Blood, 2020, 136, 2-2.	1.4	0