

Alison A Bertuch

List of Publications by Year in descending order

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Version: 2024-02-01

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	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
2	Editing <i>TINF2</i> as a potential therapeutic approach to restore telomere length in dyskeratosis congenita. Blood, 2022, 140, 608-618.	1.4	5
3	Clinical and functional characterization of telomerase variants in patients with pediatric acute myeloid leukemia/myelodysplastic syndrome. Leukemia, 2021, 35, 269-273.	7.2	4
4	Identification and characterization of novel <i>ACD</i> 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
5	Distinct genetic pathways define pre-malignant versus compensatory clonal hematopoiesis in Shwachman-Diamond syndrome. Nature Communications, 2021, 12, 1334.	12.8	103
6	Expansion of the clinical phenotype of <i>GALE</i> deficiency. American Journal of Medical Genetics, Part A, 2021, 185, 3118-3121.	1.2	8
7	Diagnostic workup for severe aplastic anemia in children: Consensus of the North American Pediatric Aplastic Anemia Consortium. American Journal of Hematology, 2021, 96, 1491-1504.	4.1	14
8	Association of unbalanced translocation der(1;7) with germline GATA2 mutations. Blood, 2021, 138, 2441-2445.	1.4	12
9	Genetic Dissection of Dyskeratosis Congenita-Causing Mutations in <i>TINF2</i> Using Human Pluripotent and Hematopoietic Stem Cell Models. Blood, 2021, 138, 2178-2178.	1.4	0
10	Hematopoiesis under telomere attrition at the single-cell resolution. Nature Communications, 2021, 12, 6850.	12.8	15
11	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
12	A study assessing the feasibility of randomization of pediatric and young adult patients between matched unrelated donor bone marrow transplantation and immune-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
13	Expansion of germline <i>RPS20</i> mutation phenotype to include Diamond-Blackfan anemia. Human Mutation, 2020, 41, 1918-1930.	2.5	13
14	Telomere dysfunction activates YAP1 to drive tissue inflammation. Nature Communications, 2020, 11, 4766.	12.8	42
15	Distinct Genetic Pathways Define Leukemia Predisposition Versus Adaptive Clonal Hematopoiesis in Shwachman-Diamond Syndrome. Blood, 2020, 136, 35-36.	1.4	0
16	Towards Identifying the Target of Autoimmunity in Aplastic Anemia. Blood, 2020, 136, 2-2.	1.4	0
17	Loss of Ku'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
18	From incomplete penetrance with normal telomere length to severe disease and telomere shortening in a family with monoallelic and biallelic <i>PARN</i> pathogenic variants. Human Mutation, 2019, 40, 2414-2429.	2.5	14

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19	Severe therapy-related toxicities after treatment for Hodgkin lymphoma due to a pathogenic TERT variant and shortened telomeres. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27779.	1.5	3
20	Immunosuppressive therapy for pediatric aplastic anemia: a North American Pediatric Aplastic Anemia Consortium study. <i>Haematologica</i> , 2019, 104, 1974-1983.	3.5	43
21	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	6.2	27
22	ClinGen Myeloid Malignancy Variant Curation Expert Panel recommendations for germline RUNX1 variants. <i>Blood Advances</i> , 2019, 3, 2962-2979.	5.2	110
23	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. <i>Genetics in Medicine</i> , 2019, 21, 663-675.	2.4	52
24	Understanding the evolving phenotype of vascular complications in telomere biology disorders. <i>Angiogenesis</i> , 2019, 22, 95-102.	7.2	45
25	<i>PSTPIP1</i> associated myeloid-related proteinemia inflammatory syndrome: A rare cause of childhood neutropenia associated with systemic inflammation and hyperzincemia. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27439.	1.5	23
26	Monozygotic twins with non-Down syndrome associated <i>MLL</i> -rearranged hematologic malignancy and megakaryoblastic differentiation. <i>Leukemia and Lymphoma</i> , 2019, 60, 1083-1086.	1.3	3
27	Cells Deficient in the Shwachman-Diamond Syndrome Protein SBDS or the Diamond-Blackfan Anemia Protein RPS19 Have Impaired Homologous Recombination. <i>Blood</i> , 2019, 134, 104-104.	1.4	0
28	Ku DNA End-Binding Activity Promotes Repair Fidelity and Influences End-Processing During Nonhomologous End-Joining in <i>Saccharomyces cerevisiae</i> . <i>Genetics</i> , 2018, 209, 115-128.	2.9	12
29	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. <i>Molecular and Cellular Biology</i> , 2018, 38, .	2.3	25
30	Structural Insights into Yeast Telomerase Recruitment to Telomeres. <i>Cell</i> , 2018, 172, 331-343.e13.	28.9	76
31	Somatic mutations and clonal hematopoiesis in congenital neutropenia. <i>Blood</i> , 2018, 131, 408-416.	1.4	91
32	Dyskeratosis Congenita and the Telomere Biology Disorders. <i>Pediatric Oncology</i> , 2018, , 111-135.	0.5	4
33	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. <i>Blood Advances</i> , 2018, 2, 1243-1249.	5.2	30
34	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. <i>Blood</i> , 2018, 132, 658-662.	1.4	9
35	Standardized high-sensitivity flow cytometry testing for paroxysmal nocturnal hemoglobinuria in children with acquired bone marrow failure disorders: A single center US study. <i>Cytometry Part B - Clinical Cytometry</i> , 2018, 94, 699-704.	1.5	7
36	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. <i>Blood</i> , 2018, 132, 5849-5849.	1.4	0

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37	The Shwachman-Diamond Syndrome Registry: Hematologic Complications. <i>Blood</i> , 2018, 132, 3871-3871.	1.4	0
38	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. <i>European Respiratory Journal</i> , 2017, 49, 1601640.	6.7	41
39	Features of Hepatitis in Hepatitis-associated Aplastic Anemia. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2017, 64, e7-e12.	1.8	32
40	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 232-245.	2.9	261
41	Somatic mutations in children with GATA2-associated myelodysplastic syndrome who lack other features of GATA2 deficiency. <i>Blood Advances</i> , 2017, 1, 443-448.	5.2	23
42	Thrombopoietin Measurement as a Key Component in the Evaluation of Pediatric Thrombocytosis. <i>Pediatric Blood and Cancer</i> , 2016, 63, 1484-1487.	1.5	10
43	Consider the workhorse: Nonhomologous end-joining in budding yeast. <i>Biochemistry and Cell Biology</i> , 2016, 94, 396-406.	2.0	56
44	Evaluation of Patients and Families With Concern for Predispositions to Hematologic Malignancies Within the Hereditary Hematologic Malignancy Clinic (HHMC). <i>Clinical Lymphoma, Myeloma and Leukemia</i> , 2016, 16, 417-428.e2.	0.4	74
45	Matched Unrelated Allogeneic Stem Cell Transplantation for Congenital Amegakaryocytic Thrombocytopenia: Texas Children's Hospital Experience. <i>Biology of Blood and Marrow Transplantation</i> , 2016, 22, S237.	2.0	1
46	Shorter Remission Telomere Length Predicts Delayed Neutrophil Recovery After Acute Myeloid Leukemia Therapy: A Report From the Children's Oncology Group. <i>Journal of Clinical Oncology</i> , 2016, 34, 3766-3772.	1.6	17
47	The molecular genetics of the telomere biology disorders. <i>RNA Biology</i> , 2016, 13, 696-706.	3.1	144
48	Tryptophan-Dependent Control of Colony Formation After DNA Damage via Sea3-Regulated TORC1 Signaling in <i>Saccharomyces cerevisiae</i> . <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 1379-1389.	1.8	4
49	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. <i>Journal of the National Cancer Institute</i> , 2015, 107, 384.	6.3	172
50	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. <i>Blood</i> , 2015, 126, 1679-1679.	1.4	1
51	Adenosine Deaminase 2 Deficiency As a Cause of Pure Red Cell Aplasia Mimicking Diamond Blackfan Anemia. <i>Blood</i> , 2015, 126, 3615-3615.	1.4	9
52	Matched Unrelated Allogeneic Stem Cell Transplantation for Patients with Congenital Amegakaryocytic Thrombocytopenia: Texas Children's Hospital Experience. <i>Blood</i> , 2015, 126, 5529-5529.	1.4	0
53	A Novel Radiosensitivity Phenotype in Shwachman-Diamond Syndrome Is Mediated By ER Stress Response. <i>Blood</i> , 2015, 126, 3618-3618.	1.4	0
54	The Principal Role of Ku in Telomere Length Maintenance Is Promotion of Est1 Association with Telomeres. <i>Genetics</i> , 2014, 197, 1123-1136.	2.9	16

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55	Compound Heterozygous CORO1A Mutations in Siblings with a Mucocutaneous-Immunodeficiency Syndrome of Epidermodysplasia Verruciformis-HPV, Molluscum Contagiosum and Granulomatous Tuberculoid Leprosy. <i>Journal of Clinical Immunology</i> , 2014, 34, 871-890.	3.8	78
56	Regulation of Ku-DNA Association by Yku70 C-terminal Tail and SUMO Modification. <i>Journal of Biological Chemistry</i> , 2014, 289, 10308-10317.	3.4	28
57	Diagnosis and treatment of pediatric acquired aplastic anemia (AAA): An initial survey of the North American Pediatric Aplastic Anemia Consortium (NAPAAC). <i>Pediatric Blood and Cancer</i> , 2014, 61, 869-874.	1.5	31
58	Structural variation and missense mutation in SBDS associated with Shwachman-Diamond syndrome. <i>BMC Medical Genetics</i> , 2014, 15, 64.	2.1	9
59	Outcome after Stem Cell Transplant in Patients with Dyskeratosis Congenita. <i>Biology of Blood and Marrow Transplantation</i> , 2014, 20, S178-S179.	2.0	0
60	A new mutant at the end: TPP1, telomeres, and BMF. <i>Blood</i> , 2014, 124, 2757-2758.	1.4	0
61	De Novo RPS20 Mutations in Diamond Blackfan Anemia. <i>Blood</i> , 2014, 124, 2667-2667.	1.4	1
62	Aplastic Anemia and Advocacy for Some Uniquely Pediatric Issues. , 2014, 11, .		0
63	TRF2 Interaction with Ku Heterotetramerization Interface Gives Insight into c-NHEJ Prevention at Human Telomeres. <i>Cell Reports</i> , 2013, 5, 194-206.	6.4	51
64	Short telomeres: from dyskeratosis congenita to sporadic aplastic anemia and malignancy. <i>Translational Research</i> , 2013, 162, 353-363.	5.0	73
65	Proposal for the clinical detection and management of patients and their family members with familial myelodysplastic syndrome/acute leukemia predisposition syndromes. <i>Leukemia and Lymphoma</i> , 2013, 54, 28-35.	1.3	88
66	A homozygous telomerase T-motif variant resulting in markedly reduced repeat addition processivity in siblings with Hoyeraal Hreidarsson syndrome. <i>Blood</i> , 2013, 121, 3586-3593.	1.4	28
67	GATA2 Mutations In Nonsyndromic Pediatric Myelodysplastic Syndrome. <i>Blood</i> , 2013, 122, 2778-2778.	1.4	1
68	Constitutional Telomerase-Associated Gene Variants In Pediatric Acute Myeloid Leukemia (AML) and In Association With Chemotherapy-Related Toxicities. <i>Blood</i> , 2013, 122, 1310-1310.	1.4	0
69	An S/T-Q cluster domain census unveils new putative targets under Tel1/Mec1 control. <i>BMC Genomics</i> , 2012, 13, 664.	2.8	19
70	Dyskeratosis congenita as a disorder of telomere maintenance. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2012, 730, 43-51.	1.0	116
71	Three novel truncating <i>TINF2</i> mutations causing severe dyskeratosis congenita in early childhood. <i>Clinical Genetics</i> , 2012, 81, 470-478.	2.0	74
72	Clinical, histopathologic, and genetic features of pediatric primary myelofibrosis—An entity different from adults. <i>American Journal of Hematology</i> , 2012, 87, 461-464.	4.1	42

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73	Constitutional Telomerase-Associated Gene Variants Associated with Chemotherapy-Related Toxicities in Pediatric Acute Myeloid Leukemia (AML). <i>Blood</i> , 2012, 120, 1403-1403.	1.4	0
74	A Homozygous Telomerase Reverse Transcriptase T Motif Variant Resulting in Markedly Reduced Telomerase Repeat Addition Processivity in Siblings with Hoyeraal Hreidarsson Syndrome. <i>Blood</i> , 2012, 120, 1272-1272.	1.4	0
75	Constitutional Telomerase-Associated Gene Variants in Pediatric Acute Myeloid Leukemia (AML). <i>Blood</i> , 2012, 120, 1408-1408.	1.4	0
76	A role for heterochromatin protein 1 ^h at human telomeres. <i>Genes and Development</i> , 2011, 25, 1807-1819.	5.9	93
77	Ku Must Load Directly onto the Chromosome End in Order to Mediate Its Telomeric Functions. <i>PLoS Genetics</i> , 2011, 7, e1002233.	3.5	35
78	High-Sensitivity Flow Cytometry Testing for Paroxysmal Nocturnal Hemoglobinuria in Children with Cytopenia: A Single Center Study. <i>Blood</i> , 2011, 118, 2398-2398.	1.4	0
79	Clinical and Molecular Characterization of Germline Telomerase Reverse Transcriptase (TERT) Variants in Children with Acute Myeloid Leukemia (AML). <i>Blood</i> , 2011, 118, 3571-3571.	1.4	0
80	Modeling growth and telomere dynamics in <i>Saccharomyces cerevisiae</i> . <i>Journal of Theoretical Biology</i> , 2010, 263, 353-359.	1.7	11
81	Abdominal undifferentiated small round cell tumor with unique translocation (X;19)(q13;q13.3). <i>Pediatric Blood and Cancer</i> , 2010, 54, 1041-1044.	1.5	4
82	The genetics and clinical manifestations of telomere biology disorders. <i>Genetics in Medicine</i> , 2010, 12, 753-764.	2.4	204
83	Measuring relative telomere length: Is tissue an issue?. <i>Aging</i> , 2010, 2, 756-757.	3.1	9
84	Primary Myelofibrosis In Children. <i>Blood</i> , 2010, 116, 3079-3079.	1.4	1
85	Ku's essential role in keeping telomeres intact. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 12217-12218.	7.1	19
86	Segregating YKU80 and TLC1 Alleles Underlying Natural Variation in Telomere Properties in Wild Yeast. <i>PLoS Genetics</i> , 2009, 5, e1000659.	3.5	46
87	The Association of yKu With Subtelomeric Core X Sequences Prevents Recombination Involving Telomeric Sequences. <i>Genetics</i> , 2009, 183, 453-467.	2.9	28
88	Infantile fibrosarcoma: Clinical and histologic responses to cytotoxic chemotherapy. <i>Pediatric Blood and Cancer</i> , 2009, 53, 23-27.	1.5	38
89	Changes Mimicking New Leptomeningeal Disease After Intensity-Modulated Radiotherapy for Medulloblastoma. <i>International Journal of Radiation Oncology Biology Physics</i> , 2009, 73, 214-221.	0.8	15
90	Poly(ADP-ribose) polymerase inhibitor ABT-888 potentiates the cytotoxic activity of temozolomide in leukemia cells: influence of mismatch repair status and O ⁶ -methylguanine-DNA methyltransferase activity. <i>Molecular Cancer Therapeutics</i> , 2009, 8, 2232-2242.	4.1	77

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91	A Single Institution Experience With Pediatric Nasopharyngeal Carcinoma: High Incidence of Toxicity Associated With Platinum-based Chemotherapy Plus IMRT. <i>Journal of Pediatric Hematology/Oncology</i> , 2007, 29, 500-505.	0.6	23
92	Distinct faces of the Ku heterodimer mediate DNA repair and telomeric functions. <i>Nature Structural and Molecular Biology</i> , 2007, 14, 301-307.	8.2	88
93	The maintenance and masking of chromosome termini. <i>Current Opinion in Cell Biology</i> , 2006, 18, 247-253.	5.4	52
94	Consequences of mutations in the non-coding RMRP RNA in cartilage-hair hypoplasia. <i>Human Molecular Genetics</i> , 2005, 14, 3723-3740.	2.9	94
95	EXO1 Contributes to Telomere Maintenance in Both Telomerase-Proficient and Telomerase-Deficient <i>Saccharomyces cerevisiae</i> . <i>Genetics</i> , 2004, 166, 1651-1659.	2.9	80
96	<i>EXO1</i> Contributes to Telomere Maintenance in Both Telomerase-Proficient and Telomerase-Deficient <i>Saccharomyces cerevisiae</i> . <i>Genetics</i> , 2004, 166, 1651-1659.	2.9	29
97	The Ku Heterodimer Performs Separable Activities at Double-Strand Breaks and Chromosome Termini. <i>Molecular and Cellular Biology</i> , 2003, 23, 8202-8215.	2.3	91
98	Which end: dissecting Ku's function at telomeres and double-strand breaks. <i>Genes and Development</i> , 2003, 17, 2347-2350.	5.9	31
99	The Way to the End Matters: The Role of Telomerase in Tumor Progression. <i>Cell Cycle</i> , 2003, 2, 36-38.	2.6	6
100	Telomeres: The Molecular Events Driving End-To-End Fusions. <i>Current Biology</i> , 2002, 12, R738-R740.	3.9	9
101	Telomeres and double-strand breaks: trying to make ends meet. <i>Trends in Cell Biology</i> , 1998, 8, 339-342.	7.9	40