Arleen D Auerbach

List of Publications by Year
in descending order

112	11,111	38742 50	30922 102
papers	citations	h-index	g-index

124	124	124	7877
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comparison of the clinical phenotype and haematological course of siblings with Fanconi anaemia. British Journal of Haematology, 2021, 193, 971-975.	2.5	6
2	The Loss of ALDH9A1 Is a Significant Source of Endogenous DNA Damage Which May be Reversed By the Inhibition of Polyamine Transport System. Blood, 2021, 138, 1108-1108.	1.4	0
3	A founder variant in the South Asian population leads to a high prevalence ofFANCLFanconi anemia cases in India. Human Mutation, 2020, 41, 122-128.	2.5	10
4	Association of clinical severity with FANCB variant type in Fanconi anemia. Blood, 2020, 135, 1588-1602.	1.4	18
5	Distinct roles of BRCA2 in replication fork protection in response to hydroxyurea and DNA interstrand cross-links. Genes and Development, 2020, 34, 832-846.	5.9	48
6	A comprehensive approach to identification of pathogenic FANCA variants in Fanconi anemia patients and their families. Human Mutation, 2018, 39, 237-254.	2.5	35
7	Somatic mosaicism of an intragenic <scp>FANCB</scp> duplication in both fibroblast and peripheral blood cells observed in a Fanconi anemia patient leads to milder phenotype. Molecular Genetics & Genomic Medicine, 2018, 6, 77-91.	1.2	28
8	Clinical Severity Correlates with in Vitro Residual Function of FANCB Missense Variants. Blood, 2018, 132, 2588-2588.	1.4	0
9	Natural history and management of <scp>F</scp>anconi anemia patients with head and neck cancer: A 10â€year followâ€up. Laryngoscope, 2016, 126, 870-879.	2.0	71
10	Paternal or Maternal Uniparental Disomy of Chromosome 16 Resulting in Homozygosity of a Mutant Allele Causes Fanconi Anemia. Human Mutation, 2016, 37, 465-468.	2.5	7
11	Diagnosis of Fanconi Anemia by Diepoxybutane Analysis. Current Protocols in Human Genetics, 2015, 85, 8.7.1-8.7.17.	3.5	48
12	Deficiency of UBE2T, the E2ÂUbiquitin Ligase Necessary for FANCD2 and FANCI Ubiquitination, Causes FA-T Subtype of Fanconi Anemia. Cell Reports, 2015, 12, 35-41.	6.4	107
13	A Dominant Mutation in Human RAD51 Reveals Its Function in DNA Interstrand Crosslink Repair Independent of Homologous Recombination. Molecular Cell, 2015, 59, 478-490.	9.7	227
14	Comprehensive Analysis of Pathogenic Deletion Variants in Fanconi Anemia Genes. Human Mutation, 2014, 35, n/a-n/a.	2.5	35
15	Regulation of multiple DNA repair pathways by the Fanconi anemia protein SLX4. Blood, 2013, 121, 54-63.	1.4	146
16	Telomere Phenotypes in Females with Heterozygous Mutations in the Dyskeratosis Congenita 1 (DKC1) Gene. Human Mutation, 2013, 34, 1481-1485.	2.5	85
17	Massively parallel sequencing, aCGH, and RNA-Seq technologies provide a comprehensive molecular diagnosis of Fanconi anemia. Blood, 2013, 121, e138-e148.	1.4	74
18	FAAP20: a novel ubiquitin-binding FA nuclear core-complex protein required for functional integrity of the FA-BRCA DNA repair pathway. Blood, 2012, 119, 3285-3294.	1.4	78

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19	Sensitive quantification of mosaicism using high density SNP arrays and the cumulative distribution function. Molecular Genetics and Metabolism, 2012, 105, 665-671.	1.1	18
20	The clinical phenotype of children with Fanconi anemia caused by biallelic FANCD1/BRCA2 mutations. Pediatric Blood and Cancer, 2012, 58, 462-465.	1.5	42
21	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	2.5	10
22	Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 790-792.	17.5	0
23	Postoperative Clinical Radiosensitivity in Patients With Fanconi Anemia and Head and Neck Squamous Cell Carcinoma. JAMA Otolaryngology, 2011, 137, 930.	1.2	49
24	Origin, functional role, and clinical impact of Fanconi anemia FANCA mutations. Blood, 2011, 117, 3759-3769.	1.4	108
25	Mutations of the SLX4 gene in Fanconi anemia. Nature Genetics, 2011, 43, 142-146.	21.4	291
26	Mutation (variation) databases and registries: a rationale for coordination of efforts. Nature Reviews Genetics, 2011, 12, 881-881.	16.3	11
27	Correct mRNA Processing at a Mutant TT Splice Donor in FANCC Ameliorates the Clinical Phenotype in Patients and Is Enhanced by Delivery of Suppressor U1 snRNAs. American Journal of Human Genetics, 2010, 87, 480-493.	6.2	59
28	FANCJ/BRIP1 recruitment and regulation of FANCD2 in DNA damage responses. Chromosoma, 2010, 119, 637-649.	2.2	22
29	Fanconi anemia and its diagnosis. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2009, 668, 4-10.	1.0	439
30	Identification and characterization of mutations in FANCL gene: A second case of Fanconi anemia belonging to FA-L complementation group. Human Mutation, 2009, 30, E761-E770.	2.5	23
31	Fludarabineâ€based cytoreductive regimen and Tâ€cellâ€depleted grafts from alternative donors for the treatment of highâ€risk patients with Fanconi anaemia. British Journal of Haematology, 2008, 140, 644-655.	2.5	79
32	FANCONI ANEMIA: A MODEL FOR GENETIC CAUSES OF ABNORMAL BRAIN DEVELOPMENT. Developmental Medicine and Child Neurology, 2008, 34, 1081-1084.	2.1	15
33	The Human Variome Project. Science, 2008, 322, 861-862.	12.6	63
34	Phenotyping Genetic Diseases Using an Extension of Âµ-Scores for Multivariate Data. Statistical Applications in Genetics and Molecular Biology, 2008, 7, Article 19.	0.6	14
35	Stem Cell Collection and Gene Transfer in Fanconi Anemia. Molecular Therapy, 2007, 15, 211-219.	8.2	166
36	Genetic Heterogeneity among Fanconi Anemia Heterozygotes and Risk of Cancer. Cancer Research, 2007, 67, 9591-9596.	0.9	102

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37	Unrelated donor bone marrow transplantation for the treatment of Fanconi anemia. Blood, 2007, 109, 2256-2262.	1.4	188
38	Hypomorphic Mutations in the Gene Encoding a Key Fanconi Anemia Protein, FANCD2, Sustain a Significant Group of FA-D2 Patients with Severe Phenotype. American Journal of Human Genetics, 2007, 80, 895-910.	6.2	115
39	Chemotherapy for myeloid malignancy in children with Fanconi anemia. Pediatric Blood and Cancer, 2007, 48, 668-672.	1.5	45
40	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. Nature Genetics, 2007, 39, 162-164.	21.4	556
41	FAAP100 is essential for activation of the Fanconi anemia-associated DNA damage response pathway. EMBO Journal, 2007, 26, 2104-2114.	7.8	130
42	FANCI is a second monoubiquitinated member of the Fanconi anemia pathway. Nature Structural and Molecular Biology, 2007, 14, 564-567.	8.2	250
43	Matched sibling donor haematopoietic stem cell transplantation in Fanconi anaemia: an update of the Cincinnati Children's experience. British Journal of Haematology, 2007, 136, 633-640.	2.5	65
44	Successful engraftment without radiation after fludarabine-based regimen in Fanconi anemia patients undergoing genotypically identical donor hematopoietic cell transplantation. Pediatric Blood and Cancer, 2006, 46, 630-636.	1.5	79
45	Reply to Dr. Strom regarding â€œA call for mutationsâ€• Genetics in Medicine, 2006, 8, 459-459.	2.4	0
46	High Incidence of Hematopoietic Stem Cell Mosaicism in Fanconi Anemia.. Blood, 2006, 108, 993-993.	1.4	14
47	GST genotype may modify clinical phenotype in patients with Fanconi anaemia. British Journal of Haematology, 2005, 131, 118-122.	2.5	12
48	The BRCA1-interacting helicase BRIP1 is deficient in Fanconi anemia. Nature Genetics, 2005, 37, 931-933.	21.4	337
49	Spectrum of sequence variations in the FANCA gene: An International Fanconi Anemia Registry (IFAR) study. Human Mutation, 2005, 25, 142-149.	2.5	71
50	A Rapid Method for Retrovirus-Mediated Identification of Complementation Groups in Fanconi Anemia Patients. Molecular Therapy, 2005, 12, 976-984.	8.2	79
51	A call for mutations. Genetics in Medicine, 2005, 7, 370-370.	2.4	5
52	Fanconi anemia in Ashkenazi Jews. Familial Cancer, 2004, 3, 241-248.	1.9	49
53	Successful umbilical cord blood transplantation for Fanconi anemia using preimplantation genetic diagnosis for HLA-matched donor. American Journal of Hematology, 2004, 77, 397-399.	4.1	43
54	Fatal Hemorrhage from Androgen-Related Hepatic Adenoma After Hematopoietic Cell Transplantation. Journal of Pediatric Hematology/Oncology, 2004, 26, 16-18.	0.6	18

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55	Germline mutations in BRCA2: shared genetic susceptibility to breast cancer, early onset leukemia, and Fanconi anemia. Blood, 2004, 103, 3226-3229.	1.4	194
56	Hematopoietic Cell Transplantation in Fanconi Anemia Patients with Biallelic BRCA2 Mutations.. Blood, 2004, 104, 2838-2838.	1.4	2
57	High Sensitivity and Specificity of Retroviral Complementation Group Assignment in Primary T Cells of Fanconi Anemia (FA) Patients.. Blood, 2004, 104, 3249-3249.	1.4	36
58	A Rapid Method for Retroviral Mediated Subtyping of Complementation Group in Fanconi Anemia Patients.. Blood, 2004, 104, 5261-5261.	1.4	1
59	Spectrum of sequence variation in theFANCG gene: An International Fanconi Anemia Registry (IFAR) study. Human Mutation, 2003, 21, 158-168.	2.5	40
60	Diagnosis of Fanconi Anemia by Diepoxybutane Analysis. Current Protocols in Human Genetics, 2003, 37, Unit 8.7.	3.5	64
61	Human Papillomavirus DNA and p53 Polymorphisms in Squamous Cell Carcinomas From Fanconi Anemia Patients. Journal of the National Cancer Institute, 2003, 95, 1718-1721.	6.3	147
62	High Incidence of Head and Neck Squamous Cell Carcinoma in Patients With Fanconi Anemia. JAMA Otolaryngology, 2003, 129, 106.	1.2	321
63	Shared Genetic Susceptibility to Breast Cancer, Brain Tumors, and Fanconi Anemia. Journal of the National Cancer Institute, 2003, 95, 1548-1551.	6.3	183
64	A 20-year perspective on the International Fanconi Anemia Registry (IFAR). Blood, 2003, 101, 1249-1256.	1.4	696
65	Phenotypic correction of primary Fanconi anemia T cells with retroviral vectors as a diagnostic tool. Experimental Hematology, 2002, 30, 410-420.	0.4	89
66	Evaluation of Growth and Hormonal Status in Patients Referred to the International Fanconi Anemia Registry. Pediatrics, 2001, 107, 744-754.	2.1	99
67	Stem cell transplantation for the treatment of Fanconi anaemia using a fludarabine-based cytoreductive regimen and T-cell-depleted related HLA-mismatched peripheral blood stem cell grafts. British Journal of Haematology, 2000, 111, 1153-1157.	2.5	41
68	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	2.9	39
69	New recessive syndrome characterized by increased chromosomal breakage and several findings which overlap with Fanconi anemia. , 1998, 78, 70-75.		7
70	Identification ofAlu-mediated deletions in the Fanconi anemia geneFAA. , 1998, 12, 145-152.		47
71	Construction of a High-Resolution Physical and Transcription Map of Chromosome 16q24.3: A Region of Frequent Loss of Heterozygosity in Sporadic Breast Cancer. Genomics, 1998, 50, 1-8.	2.9	28
72	Characterization and Screening for Mutations of the Growth Arrest-Specific 11 (GAS11) andC16orf3Genes at 16q24.3 in Breast Cancer. Genomics, 1998, 52, 325-331.	2.9	47

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73	Identification of Aluâ€mediated deletions in the Fanconi anemia gene FAA. Human Mutation, 1998, 12, 145-152.	2.5	3
74	Disorders of DNA replication and repair. Current Opinion in Pediatrics, 1997, 9, 600-616.	2.0	39
75	Fanconi Anemia: Genetic Testing in Ashkenazi Jews. Genetic Testing and Molecular Biomarkers, 1997, 1, 27-33.	1.7	23
76	The Genomic Organization of the Fanconi Anemia Group A (FAA) Gene. Genomics, 1997, 41, 309-314.	2.9	51
77	Phenotypic Consequences of Mutations in the Fanconi Anemia FAC Gene: An International Fanconi Anemia Registry Study. Blood, 1997, 90, 105-110.	1.4	119
78	New molecular diagnostic tests for two congenital forms of anemia. , 1997, 11, 17-22.		9
79	Diagnosis of Fanconi anemia in patients without congenital malformations: An international Fanconi anemia registry study. , 1997, 68, 58-61.		114
80	Interstitial lung disease in an adult with Fanconi anemia: Clues to the pathogenesis. , 1997, 69, 315-319.		2
81	Diagnosis of Fanconi anemia in patients without congenital malformations: An international Fanconi anemia registry study. American Journal of Medical Genetics Part A, 1997, 68, 58-61.	2.4	1
82	Phenotypic Consequences of Mutations in the Fanconi Anemia FAC Gene: An International Fanconi Anemia Registry Study. Blood, 1997, 90, 105-110.	1.4	7
83	Possible new variant of Nijmegen breakage syndrome. , 1996, 65, 21-26.		26
84	Positional cloning of the Fanconi anaemia group A gene. Nature Genetics, 1996, 14, 324-328.	21.4	294
85	Fanconi Anemia. Dermatologic Clinics, 1995, 13, 41-49.	1.7	53
86	Identification of Cytosolic Proteins That Bind to the Fanconi Anemia Complementation Group C Polypeptide in Vitro. Journal of Biological Chemistry, 1995, 270, 9876-9882.	3.4	52
87	A trial of recombinant human superoxide dismutase in patients with Fanconi anaemia. British Journal of Haematology, 1993, 85, 406-408.	2.5	12
88	Haemopoietic stem/progenitor cell transplant in Fanconi anaemia using HLAâ€matched sibling umbilical cord blood cells. British Journal of Haematology, 1993, 85, 419-422.	2.5	67
89	The Need for More Accurate and Timely Diagnosis in Fanconi Anemia: A Report From the International Fanconi Anemia Registry. Pediatrics, 1993, 91, 1116-1120.	2.1	158
90	Leukemia and preleukemia in Fanconi anemia. Cancer Genetics and Cytogenetics, 1992, 58, 209.	1.0	2

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91	Monozygotic twin girls with congenital malformations resembling Fanconi anemia. American Journal of Medical Genetics Part A, 1992, 42, 780-784.	2.4	19
92	Fanconi anemia: Evidence for linkage heterogeneity on chromosome 20q. Genomics, 1991, 9, 329-337.	2.9	51
93	DNA amplification for DQ typing as an adjunct to serological prenatal HLA typing for the identification of potential donors for umbilical cord blood transplantation. Human Immunology, 1991, 30, 45-49.	2.4	6
94	Fanconi anemia presenting unexpectedly in an adult kindred with no dysmorphic features. American Journal of Medicine, 1991, 91, 555-557.	1.5	26
95	Leukemia and preleukemia in Fanconi anemia patients. Cancer Genetics and Cytogenetics, 1991, 51, 1-12.	1.0	247
96	Fanconi's anaemia and pregnancy. British Journal of Haematology, 1991, 77, 410-418.	2.5	78
97	Cytogenetics in Constitutional Aplastic Anemia. , 1990, , 51-62.		1
98	Dominantly transmitted hematologic dysfunction clinically similar to fanconi's anemia. American Journal of Hematology, 1989, 32, 241-247.	4.1	2
99	Hematopoietic Reconstitution in a Patient with Fanconi's Anemia by Means of Umbilical-Cord Blood from an HLA-Identical Sibling. New England Journal of Medicine, 1989, 321, 1174-1178.	27.0	1,916
100	International Fanconi Anemia Registry: First Report. , 1989, , 3-17.		10
101	HLA typing used with cultured amniotic and chorionic villus cells for early prenatal diagnosis or parentage testing without one parent's availability. Human Immunology, 1986, 16, 200-204.	2.4	21
102	Clastogen-induced chromosomal breakage as a marker for first trimester prenatal diagnosis of Fanconi anemia. Human Genetics, 1986, 73, 86-88.	3.8	60
103	Fanconi Anemia: Prenatal Diagnosis in 30 Fetuses at Risk. Pediatrics, 1985, 76, 794-800.	2.1	70
104	Dermatologic birth defects and congenital skin disease. Journal of the American Academy of Dermatology, 1984, 11, 974-983.	1.2	2
105	Effect of procarbazine and cyclophosphamide on chromosome breakage in Fanconi anemia cells: Relevance to bone marrow transplantation. Cancer Genetics and Cytogenetics, 1983, 9, 25-36.	1.0	41
106	Acute myeloid leukemia as the first hematologic manifestation of fanconi anemia. American Journal of Hematology, 1982, 12, 289-300.	4.1	44
107	First announcement of the Fanconi anemia International Registry. Human Genetics, 1982, 61, 83-83.	3.8	1
108	Prenatal and Postnatal Diagnosis and Carrier Detection of Fanconi Anemia by a Cytogenetic Method. Pediatrics, 1981, 67, 128-135.	2.1	208

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109	Carcinogen-induced chromosome breakage in chromosome instability syndromes. Cancer Genetics and Cytogenetics, 1979, 1, 21-28.	1.0	29
110	Carcinogen-induced chromosome breakage in Fanconi's anaemia heterozygous cells. Nature, 1978, 271, 69-71.	27.8	82
111	Susceptibility of Fanconi's anaemia fibroblasts to chromosome damage by carcinogens. Nature, 1976, 261, 494-496.	27.8	321
112	Hematopoietic Cell Transplantation for Fanconi's Anemia. , 0, , 1178-1199.		0