

# Arleen D Auerbach

## List of Publications by Year in descending order

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112  
papers

11,111  
citations

38742

50  
h-index

30922

102  
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124  
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124  
docs citations

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times ranked

7877  
citing authors

#	ARTICLE	IF	CITATIONS
1	Hematopoietic Reconstitution in a Patient with Fanconi's Anemia by Means of Umbilical-Cord Blood from an HLA-Identical Sibling. <i>New England Journal of Medicine</i> , 1989, 321, 1174-1178.	27.0	1,916
2	A 20-year perspective on the International Fanconi Anemia Registry (IFAR). <i>Blood</i> , 2003, 101, 1249-1256.	1.4	696
3	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. <i>Nature Genetics</i> , 2007, 39, 162-164.	21.4	556
4	Fanconi anemia and its diagnosis. <i>Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis</i> , 2009, 668, 4-10.	1.0	439
5	The BRCA1-interacting helicase BRIP1 is deficient in Fanconi anemia. <i>Nature Genetics</i> , 2005, 37, 931-933.	21.4	337
6	Susceptibility of Fanconi's anaemia fibroblasts to chromosome damage by carcinogens. <i>Nature</i> , 1976, 261, 494-496.	27.8	321
7	High Incidence of Head and Neck Squamous Cell Carcinoma in Patients With Fanconi Anemia. <i>JAMA Otolaryngology</i> , 2003, 129, 106.	1.2	321
8	Positional cloning of the Fanconi anaemia group A gene. <i>Nature Genetics</i> , 1996, 14, 324-328.	21.4	294
9	Mutations of the SLX4 gene in Fanconi anemia. <i>Nature Genetics</i> , 2011, 43, 142-146.	21.4	291
10	FANCI is a second monoubiquitinated member of the Fanconi anemia pathway. <i>Nature Structural and Molecular Biology</i> , 2007, 14, 564-567.	8.2	250
11	Leukemia and preleukemia in Fanconi anemia patients. <i>Cancer Genetics and Cytogenetics</i> , 1991, 51, 1-12.	1.0	247
12	A Dominant Mutation in Human RAD51 Reveals Its Function in DNA Interstrand Crosslink Repair Independent of Homologous Recombination. <i>Molecular Cell</i> , 2015, 59, 478-490.	9.7	227
13	Prenatal and Postnatal Diagnosis and Carrier Detection of Fanconi Anemia by a Cytogenetic Method. <i>Pediatrics</i> , 1981, 67, 128-135.	2.1	208
14	Germline mutations in BRCA2: shared genetic susceptibility to breast cancer, early onset leukemia, and Fanconi anemia. <i>Blood</i> , 2004, 103, 3226-3229.	1.4	194
15	Unrelated donor bone marrow transplantation for the treatment of Fanconi anemia. <i>Blood</i> , 2007, 109, 2256-2262.	1.4	188
16	Shared Genetic Susceptibility to Breast Cancer, Brain Tumors, and Fanconi Anemia. <i>Journal of the National Cancer Institute</i> , 2003, 95, 1548-1551.	6.3	183
17	Stem Cell Collection and Gene Transfer in Fanconi Anemia. <i>Molecular Therapy</i> , 2007, 15, 211-219.	8.2	166
18	The Need for More Accurate and Timely Diagnosis in Fanconi Anemia: A Report From the International Fanconi Anemia Registry. <i>Pediatrics</i> , 1993, 91, 1116-1120.	2.1	158

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19	Human Papillomavirus DNA and p53 Polymorphisms in Squamous Cell Carcinomas From Fanconi Anemia Patients. <i>Journal of the National Cancer Institute</i> , 2003, 95, 1718-1721.	6.3	147
20	Regulation of multiple DNA repair pathways by the Fanconi anemia protein SLX4. <i>Blood</i> , 2013, 121, 54-63.	1.4	146
21	FAAP100 is essential for activation of the Fanconi anemia-associated DNA damage response pathway. <i>EMBO Journal</i> , 2007, 26, 2104-2114.	7.8	130
22	Phenotypic Consequences of Mutations in the Fanconi Anemia FAC Gene: An International Fanconi Anemia Registry Study. <i>Blood</i> , 1997, 90, 105-110.	1.4	119
23	Hypomorphic Mutations in the Gene Encoding a Key Fanconi Anemia Protein, FANCD2, Sustain a Significant Group of FA-D2 Patients with Severe Phenotype. <i>American Journal of Human Genetics</i> , 2007, 80, 895-910.	6.2	115
24	Diagnosis of Fanconi anemia in patients without congenital malformations: An international Fanconi anemia registry study. <i>Blood</i> , 1997, 68, 58-61.		114
25	Origin, functional role, and clinical impact of Fanconi anemia FANCA mutations. <i>Blood</i> , 2011, 117, 3759-3769.	1.4	108
26	Deficiency of UBE2T, the E2 Ubiquitin Ligase Necessary for FANCD2 and FANCI Ubiquitination, Causes FA-T Subtype of Fanconi Anemia. <i>Cell Reports</i> , 2015, 12, 35-41.	6.4	107
27	Genetic Heterogeneity among Fanconi Anemia Heterozygotes and Risk of Cancer. <i>Cancer Research</i> , 2007, 67, 9591-9596.	0.9	102
28	Evaluation of Growth and Hormonal Status in Patients Referred to the International Fanconi Anemia Registry. <i>Pediatrics</i> , 2001, 107, 744-754.	2.1	99
29	Phenotypic correction of primary Fanconi anemia T cells with retroviral vectors as a diagnostic tool. <i>Experimental Hematology</i> , 2002, 30, 410-420.	0.4	89
30	Telomere Phenotypes in Females with Heterozygous Mutations in the Dyskeratosis Congenita 1 ( <i>DKC1</i> ) Gene. <i>Human Mutation</i> , 2013, 34, 1481-1485.	2.5	85
31	Carcinogen-induced chromosome breakage in Fanconi's anaemia heterozygous cells. <i>Nature</i> , 1978, 271, 69-71.	27.8	82
32	A Rapid Method for Retrovirus-Mediated Identification of Complementation Groups in Fanconi Anemia Patients. <i>Molecular Therapy</i> , 2005, 12, 976-984.	8.2	79
33	Successful engraftment without radiation after fludarabine-based regimen in Fanconi anemia patients undergoing genotypically identical donor hematopoietic cell transplantation. <i>Pediatric Blood and Cancer</i> , 2006, 46, 630-636.	1.5	79
34	Fludarabine-based cytoreductive regimen and T cell-depleted grafts from alternative donors for the treatment of high-risk patients with Fanconi anaemia. <i>British Journal of Haematology</i> , 2008, 140, 644-655.	2.5	79
35	Fanconi's anaemia and pregnancy. <i>British Journal of Haematology</i> , 1991, 77, 410-418.	2.5	78
36	FAAP20: a novel ubiquitin-binding FA nuclear core-complex protein required for functional integrity of the FA-BRCA DNA repair pathway. <i>Blood</i> , 2012, 119, 3285-3294.	1.4	78

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37	Massively parallel sequencing, aCGH, and RNA-Seq technologies provide a comprehensive molecular diagnosis of Fanconi anemia. <i>Blood</i> , 2013, 121, e138-e148.	1.4	74
38	Spectrum of sequence variations in the FANCA gene: An International Fanconi Anemia Registry (IFAR) study. <i>Human Mutation</i> , 2005, 25, 142-149.	2.5	71
39	Natural history and management of Fanconi anemia patients with head and neck cancer: A 10-year follow-up. <i>Laryngoscope</i> , 2016, 126, 870-879.	2.0	71
40	Fanconi Anemia: Prenatal Diagnosis in 30 Fetuses at Risk. <i>Pediatrics</i> , 1985, 76, 794-800.	2.1	70
41	Haemopoietic stem/progenitor cell transplant in Fanconi anaemia using HLA-matched sibling umbilical cord blood cells. <i>British Journal of Haematology</i> , 1993, 85, 419-422.	2.5	67
42	Matched sibling donor haematopoietic stem cell transplantation in Fanconi anaemia: an update of the Cincinnati Children's experience. <i>British Journal of Haematology</i> , 2007, 136, 633-640.	2.5	65
43	Diagnosis of Fanconi Anemia by Diepoxybutane Analysis. <i>Current Protocols in Human Genetics</i> , 2003, 37, Unit 8.7.	3.5	64
44	The Human Variome Project. <i>Science</i> , 2008, 322, 861-862.	12.6	63
45	Clastogen-induced chromosomal breakage as a marker for first trimester prenatal diagnosis of Fanconi anemia. <i>Human Genetics</i> , 1986, 73, 86-88.	3.8	60
46	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. <i>American Journal of Human Genetics</i> , 2010, 87, 480-493.	6.2	59
47	Fanconi Anemia. <i>Dermatologic Clinics</i> , 1995, 13, 41-49.	1.7	53
48	Identification of Cytosolic Proteins That Bind to the Fanconi Anemia Complementation Group C Polypeptide in Vitro. <i>Journal of Biological Chemistry</i> , 1995, 270, 9876-9882.	3.4	52
49	Fanconi anemia: Evidence for linkage heterogeneity on chromosome 20q. <i>Genomics</i> , 1991, 9, 329-337.	2.9	51
50	The Genomic Organization of the Fanconi Anemia Group A (FAA) Gene. <i>Genomics</i> , 1997, 41, 309-314.	2.9	51
51	Fanconi anemia in Ashkenazi Jews. <i>Familial Cancer</i> , 2004, 3, 241-248.	1.9	49
52	Postoperative Clinical Radiosensitivity in Patients With Fanconi Anemia and Head and Neck Squamous Cell Carcinoma. <i>JAMA Otolaryngology</i> , 2011, 137, 930.	1.2	49
53	Diagnosis of Fanconi Anemia by Diepoxybutane Analysis. <i>Current Protocols in Human Genetics</i> , 2015, 85, 8.7.1-8.7.17.	3.5	48
54	Distinct roles of BRCA2 in replication fork protection in response to hydroxyurea and DNA interstrand cross-links. <i>Genes and Development</i> , 2020, 34, 832-846.	5.9	48

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55	Identification ofAlu-mediated deletions in the Fanconi anemia geneFAA. , 1998, 12, 145-152.		47
56	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
57	Chemotherapy for myeloid malignancy in children with Fanconi anemia. Pediatric Blood and Cancer, 2007, 48, 668-672.	1.5	45
58	Acute myeloid leukemia as the first hematologic manifestation of fanconi anemia. American Journal of Hematology, 1982, 12, 289-300.	4.1	44
59	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
60	The clinical phenotype of children with Fanconi anemia caused by biallelic <i>FANCD1/BRCA2</i> mutations. Pediatric Blood and Cancer, 2012, 58, 462-465.	1.5	42
61	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
62	Stem cell transplantation for the treatment of Fanconi anaemia using a fludarabine-based cyoreductive regimen and T-cell-depleted related HLA-mismatched peripheral blood stem cell grafts. British Journal of Haematology, 2000, 111, 1153-1157.	2.5	41
63	Spectrum of sequence variation in theFANCG gene: An International Fanconi Anemia Registry (IFAR) study. Human Mutation, 2003, 21, 158-168.	2.5	40
64	Disorders of DNA replication and repair. Current Opinion in Pediatrics, 1997, 9, 600-616.	2.0	39
65	ThePISSLREGene: Structure, Exon Skipping, and Exclusion as Tumor Suppressor in Breast Cancer. Genomics, 1999, 56, 90-97.	2.9	39
66	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
67	Comprehensive Analysis of Pathogenic Deletion Variants in Fanconi Anemia Genes. Human Mutation, 2014, 35, n/a-n/a.	2.5	35
68	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
69	Carcinogen-induced chromosome breakage in chromosome instability syndromes. Cancer Genetics and Cytogenetics, 1979, 1, 21-28.	1.0	29
70	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
71	Somatic mosaicism of an intragenic <i>FANCB</i> 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
72	Fanconi anemia presenting unexpectedly in an adult kindred with no dysmorphic features. American Journal of Medicine, 1991, 91, 555-557.	1.5	26

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73	Possible new variant of Nijmegen breakage syndrome. , 1996, 65, 21-26.		26
74	Fanconi Anemia: Genetic Testing in Ashkenazi Jews. Genetic Testing and Molecular Biomarkers, 1997, 1, 27-33.	1.7	23
75	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
76	FANCI/BRIP1 recruitment and regulation of FANCD2 in DNA damage responses. Chromosoma, 2010, 119, 637-649.	2.2	22
77	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
78	Monozygotic twin girls with congenital malformations resembling Fanconi anemia. American Journal of Medical Genetics Part A, 1992, 42, 780-784.	2.4	19
79	Fatal Hemorrhage from Androgen-Related Hepatic Adenoma After Hematopoietic Cell Transplantation. Journal of Pediatric Hematology/Oncology, 2004, 26, 16-18.	0.6	18
80	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
81	Association of clinical severity with FANCB variant type in Fanconi anemia. Blood, 2020, 135, 1588-1602.	1.4	18
82	FANCONI ANEMIA: A MODEL FOR GENETIC CAUSES OF ABNORMAL BRAIN DEVELOPMENT. Developmental Medicine and Child Neurology, 2008, 34, 1081-1084.	2.1	15
83	Phenotyping Genetic Diseases Using an Extension of $\hat{\mu}$ -Scores for Multivariate Data. Statistical Applications in Genetics and Molecular Biology, 2008, 7, Article 19.	0.6	14
84	High Incidence of Hematopoietic Stem Cell Mosaicism in Fanconi Anemia.. Blood, 2006, 108, 993-993.	1.4	14
85	A trial of recombinant human superoxide dismutase in patients with Fanconi anaemia. British Journal of Haematology, 1993, 85, 406-408.	2.5	12
86	GST genotype may modify clinical phenotype in patients with Fanconi anaemia. British Journal of Haematology, 2005, 131, 118-122.	2.5	12
87	Mutation (variation) databases and registries: a rationale for coordination of efforts. Nature Reviews Genetics, 2011, 12, 881-881.	16.3	11
88	Human variome project country nodes: Documenting genetic information within a country. Human Mutation, 2012, 33, 1513-1519.	2.5	10
89	A founder variant in the South Asian population leads to a high prevalence of <i>FANCL</i> Fanconi anemia cases in India. Human Mutation, 2020, 41, 122-128.	2.5	10
90	International Fanconi Anemia Registry: First Report. , 1989, , 3-17.		10

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91	New molecular diagnostic tests for two congenital forms of anemia. , 1997, 11, 17-22.		9
92	New recessive syndrome characterized by increased chromosomal breakage and several findings which overlap with Fanconi anemia. , 1998, 78, 70-75.		7
93	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
94	Phenotypic Consequences of Mutations in the Fanconi Anemia FAC Gene: An International Fanconi Anemia Registry Study. Blood, 1997, 90, 105-110.	1.4	7
95	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
96	Comparison of the clinical phenotype and haematological course of siblings with Fanconi anaemia. British Journal of Haematology, 2021, 193, 971-975.	2.5	6
97	A call for mutations. Genetics in Medicine, 2005, 7, 370-370.	2.4	5
98	Identification of Alu-mediated deletions in the Fanconi anemia gene FAA. Human Mutation, 1998, 12, 145-152.	2.5	3
99	Dermatologic birth defects and congenital skin disease. Journal of the American Academy of Dermatology, 1984, 11, 974-983.	1.2	2
100	Dominantly transmitted hematologic dysfunction clinically similar to fanconi's anemia. American Journal of Hematology, 1989, 32, 241-247.	4.1	2
101	Leukemia and preleukemia in Fanconi anemia. Cancer Genetics and Cytogenetics, 1992, 58, 209.	1.0	2
102	Interstitial lung disease in an adult with Fanconi anemia: Clues to the pathogenesis. , 1997, 69, 315-319.		2
103	Hematopoietic Cell Transplantation in Fanconi Anemia Patients with Biallelic BRCA2 Mutations.. Blood, 2004, 104, 2838-2838.	1.4	2
104	First announcement of the Fanconi anemia International Registry. Human Genetics, 1982, 61, 83-83.	3.8	1
105	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
106	A Rapid Method for Retroviral Mediated Subtyping of Complementation Group in Fanconi Anemia Patients.. Blood, 2004, 104, 5261-5261.	1.4	1
107	Cytogenetics in Constitutional Aplastic Anemia. , 1990, , 51-62.		1
108	Reply to Dr. Strom regarding "A call for mutations". Genetics in Medicine, 2006, 8, 459-459.	2.4	0

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109	Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 790-792.	17.5	0
110	Clinical Severity Correlates with in Vitro Residual Function of FANCB Missense Variants. Blood, 2018, 132, 2588-2588.	1.4	0
111	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
112	Hematopoietic Cell Transplantation for Fanconi's Anemia. , 0, , 1178-1199.		0