

Blanche P Alter

List of Publications by Year in descending order

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207
papers

11,819
citations

28190

55
h-index

30848

102
g-index

211
all docs

211
docs citations

211
times ranked

9575
citing authors

#	ARTICLE	IF	CITATIONS
1	Pathophysiology and management of inherited bone marrow failure syndromes. <i>Blood Reviews</i> , 2010, 24, 101-122.	2.8	432
2	Cancer incidence in persons with Fanconi anemia. <i>Blood</i> , 2003, 101, 822-826.	0.6	430
3	Cancer in dyskeratosis congenita. <i>Blood</i> , 2009, 113, 6549-6557.	0.6	413
4	Diagnosing and treating Diamond Blackfan anaemia: results of an international clinical consensus conference. <i>British Journal of Haematology</i> , 2008, 142, 859-876.	1.2	408
5	The incidence of leukemia and mortality from sepsis in patients with severe congenital neutropenia receiving long-term G-CSF therapy. <i>Blood</i> , 2006, 107, 4628-4635.	0.6	394
6	Cancer in Fanconi anemia, 1927-2001. <i>Cancer</i> , 2003, 97, 425-440.	2.0	387
7	TINF2, a Component of the Shelterin Telomere Protection Complex, Is Mutated in Dyskeratosis Congenita. <i>American Journal of Human Genetics</i> , 2008, 82, 501-509.	2.6	368
8	Very short telomere length by flow fluorescence in situ hybridization identifies patients with dyskeratosis congenita. <i>Blood</i> , 2007, 110, 1439-1447.	0.6	296
9	Cancer Survivorship—Genetic Susceptibility and Second Primary Cancers: Research Strategies and Recommendations. <i>Journal of the National Cancer Institute</i> , 2006, 98, 15-25.	3.0	295
10	Malignancies and survival patterns in the National Cancer Institute inherited bone marrow failure syndromes cohort study. <i>British Journal of Haematology</i> , 2010, 150, 179-188.	1.2	272
11	Incidence of neoplasia in Diamond Blackfan anemia: a report from the Diamond Blackfan Anemia Registry. <i>Blood</i> , 2012, 119, 3815-3819.	0.6	263
12	Risk of head and neck squamous cell cancer and death in patients with Fanconi anemia who did and did not receive transplants. <i>Blood</i> , 2005, 105, 67-73.	0.6	244
13	Cancer in the National Cancer Institute inherited bone marrow failure syndrome cohort after fifteen years of follow-up. <i>Haematologica</i> , 2018, 103, 30-39.	1.7	236
14	Fanconi's anemia and malignancies. , 1996, 53, 99-110.		225
15	Telomere shortening and loss of self-renewal in dyskeratosis congenita induced pluripotent stem cells. <i>Nature</i> , 2011, 474, 399-402.	13.7	220
16	Clinical and molecular features associated with biallelic mutations in FANCD1/BRCA2. <i>Journal of Medical Genetics</i> , 2006, 44, 1-9.	1.5	215
17	Disruption of telomerase trafficking by TCAB1 mutation causes dyskeratosis congenita. <i>Genes and Development</i> , 2011, 25, 11-16.	2.7	213
18	Stable long-term risk of leukaemia in patients with severe congenital neutropenia maintained on G-CSF therapy. <i>British Journal of Haematology</i> , 2010, 150, 196-199.	1.2	211

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19	Androgens and liver tumors: Fanconi's anemia and non-Fanconi's conditions. <i>American Journal of Hematology</i> , 2004, 77, 257-267.	2.0	207
20	Germline mutations of regulator of telomere elongation helicase 1, RTEL1, in Dyskeratosis congenita. <i>Human Genetics</i> , 2013, 132, 473-480.	1.8	198
21	Dyskeratosis Congenita. <i>Hematology/Oncology Clinics of North America</i> , 2009, 23, 215-231.	0.9	195
22	Telomere length is associated with disease severity and declines with age in dyskeratosis congenita. <i>Haematologica</i> , 2012, 97, 353-359.	1.7	194
23	Cancer risks in Fanconi anemia: findings from the German Fanconi Anemia Registry. <i>Haematologica</i> , 2008, 93, 511-517.	1.7	182
24	Fanconi Anemia. <i>Seminars in Hematology</i> , 2006, 43, 147-156.	1.8	174
25	Diagnosis, Genetics, and Management of Inherited Bone Marrow Failure Syndromes. <i>Hematology American Society of Hematology Education Program</i> , 2007, 2007, 29-39.	0.9	174
26	Fanconi anemia and the development of leukemia. <i>Best Practice and Research in Clinical Haematology</i> , 2014, 27, 214-221.	0.7	166
27	Comparative genome analysis delimits a chromosomal domain and identifies key regulatory elements in the alpha globin cluster. <i>Human Molecular Genetics</i> , 2001, 10, 371-382.	1.4	151
28	Cancer in Fanconi anemia. <i>Blood</i> , 2003, 101, 2072-2072.	0.6	147
29	Endocrine Abnormalities in Patients with Fanconi Anemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 2624-2631.	1.8	120
30	Telomere length in blood, buccal cells, and fibroblasts from patients with inherited bone marrow failure syndromes. <i>Aging</i> , 2010, 2, 867-874.	1.4	120
31	Genotype-phenotype associations in Fanconi anemia: A literature review. <i>Blood Reviews</i> , 2019, 37, 100589.	2.8	116
32	Outcomes of Allogeneic Hematopoietic Cell Transplantation in Patients with Dyskeratosis Congenita. <i>Biology of Blood and Marrow Transplantation</i> , 2013, 19, 1238-1243.	2.0	108
33	A Recessive Founder Mutation in Regulator of Telomere Elongation Helicase 1, RTEL1, Underlies Severe Immunodeficiency and Features of Hoyeraal Hreidarsson Syndrome. <i>PLoS Genetics</i> , 2013, 9, e1003695.	1.5	106
34	Hoyeraal-Hreidarsson syndrome caused by a germline mutation in the TEL patch of the telomere protein TPP1. <i>Genes and Development</i> , 2014, 28, 2090-2102.	2.7	106
35	Radiosensitivity in Fanconi's anemia patients. <i>Radiotherapy and Oncology</i> , 2002, 62, 345-347.	0.3	105
36	Fanconi Anemia. <i>Cancer Genetics and Cytogenetics</i> , 2000, 117, 125-131.	1.0	99

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37	How high are carrier frequencies of rare recessive syndromes? Contemporary estimates for Fanconi Anemia in the United States and Israel. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1877-1883.	0.7	95
38	Response to androgen therapy in patients with dyskeratosis congenita. <i>British Journal of Haematology</i> , 2014, 165, 349-357.	1.2	89
39	Drug discovery for Diamond-Blackfan anemia using reprogrammed hematopoietic progenitors. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	87
40	Individualized risks of first adverse events in patients with Fanconi anemia. <i>Blood</i> , 2004, 104, 350-355.	0.6	84
41	Dyskeratosis Congenita. <i>Journal of Pediatric Hematology/Oncology</i> , 1992, 14, 297-304.	0.3	82
42	Shwachman-Diamond syndrome: Report from an international conference. <i>Journal of Pediatrics</i> , 2002, 141, 266-270.	0.9	81
43	Whole-exome sequencing and functional studies identify RPS29 as a novel gene mutated in multicase Diamond-Blackfan anemia families. <i>Blood</i> , 2014, 124, 24-32.	0.6	79
44	Fanconi's anaemia and pregnancy. <i>British Journal of Haematology</i> , 1991, 77, 410-418.	1.2	78
45	Erythrocyte adenosine deaminase: diagnostic value for Diamond-Blackfan anaemia. <i>British Journal of Haematology</i> , 2013, 160, 547-554.	1.2	76
46	Endocrine Disorders in Fanconi Anemia: Recommendations for Screening and Treatment. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, 803-811.	1.8	76
47	Clinical features and outcomes of patients with Shwachman-Diamond syndrome and myelodysplastic syndrome or acute myeloid leukaemia: a multicentre, retrospective, cohort study. <i>Lancet Haematology</i> , 2020, 7, e238-e246.	2.2	73
48	Ocular and Orbital Manifestations of the Inherited Bone Marrow Failure Syndromes: Fanconi Anemia and Dyskeratosis Congenita. <i>Ophthalmology</i> , 2010, 117, 615-622.	2.5	72
49	Novel and known ribosomal causes of Diamond-Blackfan anaemia identified through comprehensive genomic characterisation. <i>Journal of Medical Genetics</i> , 2017, 54, 417-425.	1.5	71
50	Dyskeratosis congenita: The first NIH clinical research workshop. <i>Pediatric Blood and Cancer</i> , 2009, 53, 520-523.	0.8	66
51	The role of telomere biology in bone marrow failure and other disorders. <i>Mechanisms of Ageing and Development</i> , 2008, 129, 35-47.	2.2	64
52	Increased risk of colon cancer and osteogenic sarcoma in Diamond-Blackfan anemia. <i>Blood</i> , 2018, 132, 2205-2208.	0.6	64
53	Squamous cell carcinomas in patients with Fanconi anemia and dyskeratosis congenita: A search for human papillomavirus. <i>International Journal of Cancer</i> , 2013, 133, 1513-1515.	2.3	63
54	Telomere length in inherited bone marrow failure syndromes. <i>Haematologica</i> , 2015, 100, 49-54.	1.7	63

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55	Frequency and natural history of inherited bone marrow failure syndromes: the Israeli Inherited Bone Marrow Failure Registry. <i>Haematologica</i> , 2010, 95, 1300-1307.	1.7	57
56	Inherited bone marrow failure syndromes: considerations pre- and posttransplant. <i>Blood</i> , 2017, 130, 2257-2264.	0.6	57
57	Lung transplantation for pulmonary fibrosis in dyskeratosis congenita: Case Report and systematic literature review. <i>BMC Blood Disorders</i> , 2011, 11, 3.	0.9	56
58	Fanconi Anemia. <i>JAMA Otolaryngology</i> , 2005, 131, 635.	1.5	55
59	Bone marrow failure syndromes in children. <i>Pediatric Clinics of North America</i> , 2002, 49, 973-988.	0.9	54
60	A comprehensive functional characterization of BRCA2 variants associated with Fanconi anemia using mouse ES cell-based assay. <i>Blood</i> , 2011, 118, 2430-2442.	0.6	53
61	p53 Protein Overexpression in Bone Marrow Biopsies of Patients With Shwachman-Diamond Syndrome Has a Prevalence Similar to That of Patients With Refractory Anemia. <i>Archives of Pathology and Laboratory Medicine</i> , 2002, 126, 452-455.	1.2	51
62	Comparison of Chromosome Breakage in Non-Mosaic and Mosaic Patients with Fanconi Anemia, Relatives, and Patients with Other Inherited Bone Marrow Failure Syndromes. <i>Cytogenetic and Genome Research</i> , 2014, 144, 15-27.	0.6	47
63	Understanding the evolving phenotype of vascular complications in telomere biology disorders. <i>Angiogenesis</i> , 2019, 22, 95-102.	3.7	45
64	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. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 1422-1428.	2.0	43
65	Splenic peliosis and rupture in patients with dyskeratosis congenita on androgens and granulocyte colony-stimulating factor. <i>British Journal of Haematology</i> , 2007, 138, 815-817.	1.2	42
66	Correlation of Leukocyte Telomere Length Measurement Methods in Patients with Dyskeratosis Congenita and in Their Unaffected Relatives. <i>International Journal of Molecular Sciences</i> , 2017, 18, 1765.	1.8	42
67	Re-equilibration of imbalanced NAD metabolism ameliorates the impact of telomere dysfunction. <i>EMBO Journal</i> , 2020, 39, e103420.	3.5	42
68	Pulmonary arteriovenous malformations: an uncharacterised phenotype of dyskeratosis congenita and related telomere biology disorders. <i>European Respiratory Journal</i> , 2017, 49, 1601640.	3.1	41
69	Fanconi's Anemia, transplantation, and cancer. <i>Pediatric Transplantation</i> , 2005, 9, 81-86.	0.5	38
70	Thinking of VACTERL? Rule out Fanconi Anemia according to PHENOS. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1520-1524.	0.7	38
71	Clonal chromosomal abnormalities in Fanconi's anaemia: what do they really mean?. <i>British Journal of Haematology</i> , 1993, 85, 627-630.	1.2	36
72	Inherited bone marrow failure syndromes: considerations pre- and posttransplant. <i>Hematology American Society of Hematology Education Program</i> , 2017, 2017, 88-95.	0.9	36

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73	JUVENILE CHRONIC MYELOGENOUS LEUKAEMIA: THE ONLY EXAMPLE OF TRULY FETAL (NOT FETAL-LIKE) ERYTHROPOIESIS. <i>British Journal of Haematology</i> , 1990, 76, 307-310.	1.2	35
74	Immune status of patients with inherited bone marrow failure syndromes. <i>American Journal of Hematology</i> , 2015, 90, 702-708.	2.0	34
75	Disease progression and clinical outcomes in telomere biology disorders. <i>Blood</i> , 2022, 139, 1807-1819.	0.6	34
76	The Second Pediatric Blood and Marrow Transplant Consortium International Consensus Conference on Late Effects after Pediatric Hematopoietic Cell Transplantation: Defining the Unique Late Effects of Children Undergoing Hematopoietic Cell Transplantation for Immune Deficiencies, Inherited Marrow Failure Disorders, and Hemoglobinopathies. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 24-29.	2.0	33
77	Intensive Immunosuppression Therapy for Aplastic Anemia Associated with Dyskeratosis Congenita. <i>International Journal of Hematology</i> , 2006, 83, 275-276.	0.7	32
78	Bone marrow skeletal stem/progenitor cell defects in dyskeratosis congenita and telomere biology disorders. <i>Blood</i> , 2015, 125, 793-802.	0.6	31
79	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. <i>Biology of Blood and Marrow Transplantation</i> , 2017, 23, 726-735.	2.0	31
80	Patients with Fanconi anemia and AML have different cytogenetic clones than de novo cases of AML. <i>Pediatric Blood and Cancer</i> , 2012, 59, 922-924.	0.8	30
81	Similar telomere attrition rates in androgen-treated and untreated patients with dyskeratosis congenita. <i>Blood Advances</i> , 2018, 2, 1243-1249.	2.5	30
82	Treatment of dyskeratosis congenita with Granulocyte Colony-Stimulating Factor and Erythropoietin. <i>British Journal of Haematology</i> , 1997, 97, 309-311.	1.2	29
83	Sources of Uncertainty and Their Association with Medical Decision Making: Exploring Mechanisms in Fanconi Anemia. <i>Annals of Behavioral Medicine</i> , 2013, 46, 204-216.	1.7	29
84	Dubowitz Syndrome Is a Complex Comprised of Multiple, Genetically Distinct and Phenotypically Overlapping Disorders. <i>PLoS ONE</i> , 2014, 9, e98686.	1.1	29
85	Anti-Müllerian Hormone Deficiency in Females With Fanconi Anemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, 1608-1614.	1.8	29
86	Hoyeraal-Hreidarsson Syndrome due to PARN Mutations: Fourteen Years of Follow-Up. <i>Pediatric Neurology</i> , 2016, 56, 62-68.e1.	1.0	29
87	The relationship between DNA methylation and telomere length in dyskeratosis congenita. <i>Ageing Cell</i> , 2012, 11, 24-28.	3.0	28
88	Globin Chain Electrophoresis for Prenatal Diagnosis of β^2 Thalassemia. <i>Hemoglobin</i> , 1981, 5, 357-370.	0.4	27
89	Erythropoiesis is Distinct at Each Stage of Ontogeny. <i>Pediatric Research</i> , 1992, 31, 170-175.	1.1	27
90	CURRENT DIAGNOSIS OF INHERITED BONE MARROW FAILURE SYNDROMES. <i>Pediatric Hematology and Oncology</i> , 2007, 24, 87-99.	0.3	27

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91	Pregnancy in bone marrow failure syndromes: Diamond-Blackfan anaemia and Shwachman-Diamond syndrome. <i>British Journal of Haematology</i> , 1999, 107, 49-54.	1.2	26
92	The accumulation and not the specific activity of telomerase ribonucleoprotein determines telomere maintenance deficiency in X-linked dyskeratosis congenita. <i>Human Molecular Genetics</i> , 2012, 21, 721-729.	1.4	26
93	Neuropsychiatric Conditions Among Patients with Dyskeratosis Congenita: A Link with Telomere Biology?. <i>Psychosomatics</i> , 2012, 53, 230-235.	2.5	26
94	Calmodulin inhibitors improve erythropoiesis in Diamond-Blackfan anemia. <i>Science Translational Medicine</i> , 2020, 12, .	5.8	26
95	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. <i>Blood Cells, Molecules, and Diseases</i> , 2006, 37, 134-136.	0.6	25
96	Novel <i>FANCI</i> mutations in Fanconi anemia with VACTERL association. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 386-391.	0.7	25
97	Germline mutation of <i>MDM4</i> , a major p53 regulator, in a familial syndrome of defective telomere maintenance. <i>Science Advances</i> , 2020, 6, eaay3511.	4.7	25
98	Research participant interest in primary, secondary, and incidental genomic findings. <i>Genetics in Medicine</i> , 2016, 18, 1218-1225.	1.1	24
99	Beyond the triad: Inheritance, mucocutaneous phenotype, and mortality in a cohort of patients with dyskeratosis congenita. <i>Journal of the American Academy of Dermatology</i> , 2018, 78, 804-806.	0.6	23
100	Preemptive Bone Marrow Transplantation and Event-Free Survival in Fanconi Anemia. <i>Biology of Blood and Marrow Transplantation</i> , 2016, 22, 1888-1892.	2.0	22
101	Investigation of chromosome X inactivation and clinical phenotypes in female carriers of DKC1 mutations. <i>American Journal of Hematology</i> , 2016, 91, 1215-1220.	2.0	22
102	Genetic regulation of fetal haemoglobin in inherited bone marrow failure syndromes. <i>British Journal of Haematology</i> , 2013, 162, 542-546.	1.2	21
103	The limitations of qPCR telomere length measurement in diagnosing dyskeratosis congenita. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 475-479.	0.6	20
104	CNS manifestations in patients with telomere biology disorders. <i>Neurology: Genetics</i> , 2019, 5, 370.	0.9	17
105	Aplastic Anemia, Pediatric Aspects. <i>Oncologist</i> , 1996, 1, 361-366.	1.9	16
106	Bone Marrow Failure: A Child Is Not Just a Small Adult (But an Adult Can Have a Childhood Disease). <i>Hematology American Society of Hematology Education Program</i> , 2005, 2005, 96-103.	0.9	16
107	Bone marrow cell cycle markers in inherited bone marrow failure syndromes. <i>Leukemia Research</i> , 2008, 32, 1793-1799.	0.4	15
108	PROLIFERATIVE RETINOPATHY AS A COMPLICATION OF DYSKERATOSIS CONGENITA. <i>Retinal Cases and Brief Reports</i> , 2009, 3, 259-262.	0.3	15

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109	Cytokine production by bone marrow mononuclear cells in inherited bone marrow failure syndromes. <i>British Journal of Haematology</i> , 2013, 163, 81-92.	1.2	15
110	Genetic Information-Seeking Behaviors and Knowledge among Family Members and Patients with Inherited Bone Marrow Failure Syndromes. <i>Journal of Genetic Counseling</i> , 2015, 24, 760-770.	0.9	15
111	Pregnancies in patients with inherited bone marrow failure syndromes in the NCI cohort. <i>Blood</i> , 2017, 130, 1674-1676.	0.6	15
112	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. <i>Genome Medicine</i> , 2018, 10, 99.	3.6	15
113	Reduced Serum Levels of Anti-M β 1/4llergic Hormone in Females With Inherited Bone Marrow Failure Syndromes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E197-E203.	1.8	14
114	Preemptive Bone Marrow Transplantation for FANCD1/BRCA2. <i>Biology of Blood and Marrow Transplantation</i> , 2015, 21, 1796-1801.	2.0	14
115	Subsequent neoplasms and late mortality in children undergoing allogeneic transplantation for nonmalignant diseases. <i>Blood Advances</i> , 2020, 4, 2084-2094.	2.5	14
116	Gastrointestinal Hemorrhage: A Manifestation of the Telomere Biology Disorders. <i>Journal of Pediatrics</i> , 2021, 230, 55-61.e4.	0.9	14
117	PRENATAL DIAGNOSIS OF HEMOGLOBINOPATHIES: THE NEW ENGLAND APPROACH. <i>Annals of the New York Academy of Sciences</i> , 1980, 344, 151-164.	1.8	13
118	Antibody response to human papillomavirus vaccine in subjects with inherited bone marrow failure syndromes. <i>Vaccine</i> , 2014, 32, 1169-1173.	1.7	13
119	Prognostic significance of pulmonary function tests in dyskeratosis congenita, a telomere biology disorder. <i>ERJ Open Research</i> , 2019, 5, 00209-2019.	1.1	13
120	Expansion of germline <i>RPS20</i> mutation phenotype to include Diamond-Blackfan anemia. <i>Human Mutation</i> , 2020, 41, 1918-1930.	1.1	13
121	The effect of hemin in vitro and in vivo on human erythroid progenitor cells. <i>International Journal of Cell Cloning</i> , 1986, 4, 432-446.	1.6	12
122	Prenatal Diagnosis of Hemoglobinopathies: A Potential Application of High Performance Liquid Chromatography. <i>Hemoglobin</i> , 1987, 11, 341-352.	0.4	12
123	Sickle and Thalassemic Erythroid Progenitor Cells are Different from Normal. <i>Hemoglobin</i> , 1992, 16, 447-467.	0.4	12
124	The association between FANCD1/BRCA2 mutations and leukaemia. <i>British Journal of Haematology</i> , 2006, 133, 446-448.	1.2	12
125	Sequence analysis of the shelterin telomere protection complex genes in dyskeratosis congenita. <i>Journal of Medical Genetics</i> , 2011, 48, 285-288.	1.5	12
126	Estimation of the prevalence of Fanconi anemia among patients with de novo acute myelogenous leukemia who have poor recovery from chemotherapy. <i>Leukemia Research</i> , 2012, 36, 29-31.	0.4	12

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127	Genotype-phenotype association and variant characterization in Diamond-Blackfan anemia caused by pathogenic variants in <i>RPL35A</i> . <i>Haematologica</i> , 2021, 106, 1303-1310.	1.7	12
128	Pregnancy outcomes in mothers of offspring with inherited bone marrow failure syndromes. <i>Pediatric Blood and Cancer</i> , 2018, 65, e26757.	0.8	11
129	Prenatal Diagnosis: General Introduction, Methodology, and Review. <i>Hemoglobin</i> , 1988, 12, 763-772.	0.4	10
130	Otologic manifestations of Fanconi anemia and other inherited bone marrow failure syndromes. <i>Pediatric Blood and Cancer</i> , 2016, 63, 2139-2145.	0.8	10
131	Bone mineral density in patients with inherited bone marrow failure syndromes. <i>Pediatric Research</i> , 2017, 82, 458-464.	1.1	10
132	Genotype-phenotype and outcome associations in patients with Fanconi anemia: the National Cancer Institute cohort. <i>Haematologica</i> , 2023, 108, 69-82.	1.7	10
133	All in the family: Disclosure of "unwanted" information to an adolescent to benefit a relative. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2719-2724.	0.7	9
134	Pearson syndrome in a Diamond-Blackfan anemia cohort. <i>Blood</i> , 2014, 124, 312-313.	0.6	9
135	Second Allogeneic Hematopoietic Cell Transplantation for Patients with Fanconi Anemia and Bone Marrow Failure. <i>Biology of Blood and Marrow Transplantation</i> , 2015, 21, 1790-1795.	2.0	9
136	Chromosomal Aberrations and Survival after Unrelated Donor Hematopoietic Stem Cell Transplant in Patients with Fanconi Anemia. <i>Biology of Blood and Marrow Transplantation</i> , 2018, 24, 2003-2008.	2.0	9
137	Progressive reticulate skin pigmentation and anonychia in a patient with bone marrow failure. <i>Journal of the American Academy of Dermatology</i> , 2017, 77, 1194-1198.	0.6	8
138	Neutrophil functions in patients with inherited bone marrow failure syndromes. <i>Pediatric Blood and Cancer</i> , 2011, 57, 306-309.	0.8	7
139	The use of haematopoietic stem cell transplantation in Fanconi anaemia patients: a survey of decision making among families in the US and Canada. <i>Health Expectations</i> , 2015, 18, 929-941.	1.1	7
140	Complex phenotype of dyskeratosis congenita and mood dysregulation with novel homozygous <i>RTKL1</i> and <i>TPH1</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1432-1437.	0.7	7
141	Genotype-cancer association in patients with Fanconi anemia due to pathogenic variants in <i>FANCD1</i> (<i>BRCA2</i>) or <i>FANCN</i> (<i>PALB2</i>). <i>Cancer Genetics</i> , 2021, 258-259, 101-109.	0.2	7
142	Lack of effect of corticosteroids in <i>W/W^v</i> and <i>Sl/Sl^d</i> mice: These strains are not a model for steroid-responsive Diamond-Blackfan anemia. <i>European Journal of Haematology</i> , 1993, 50, 275-278.	1.1	6
143	Population Frequency of Fanconi Pathway Gene Variants and Their Association with Survival After Hematopoietic Cell Transplantation for Severe Aplastic Anemia. <i>Biology of Blood and Marrow Transplantation</i> , 2020, 26, 817-822.	2.0	6
144	Bone marrow transplantation in two multiply transfused patients with thalassaemia major. <i>British Journal of Haematology</i> , 1986, 63, 445-456.	1.2	5

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145	The CCC System: Is It Really the Answer to Pediatric MDS?. Journal of Pediatric Hematology/Oncology, 2003, 25, 426-427.	0.3	5
146	Growth hormone and the risk of malignancy. Pediatric Blood and Cancer, 2004, 43, 534-535.	0.8	5
147	Wilms tumor, AML, and medulloblastoma in a child with cancer prone syndrome of total premature chromatid separation and Fanconi anemia. Pediatric Blood and Cancer, 2010, 54, 488-488.	0.8	5
148	Gynaecological and reproductive health of women with telomere biology disorders. British Journal of Haematology, 2021, 193, 1238-1246.	1.2	5
149	Pathogenic germline <i>KZF1</i> variant alters hematopoietic gene expression profiles. Journal of Physical Education and Sports Management, 2021, 7, a006015.	0.5	5
150	Fanconi's anemia and malignancies. American Journal of Hematology, 1996, 53, 99-110.	2.0	5
151	Risk of cancer in heterozygous relatives of patients with Fanconi anemia. Genetics in Medicine, 2022, 24, 245-250.	1.1	5
152	Shwachman Diamond syndrome: narrow genotypic spectrum and variable clinical features. Pediatric Research, 2022, , .	1.1	5
153	Bone marrow failure syndromes. , 0, , 47-64.		4
154	1q21.1 deletion and a rare functional polymorphism in siblings with thrombocytopenia-absent radius-like phenotypes. Journal of Physical Education and Sports Management, 2019, 5, a004564.	0.5	4
155	Correlation of Telomere Length in Blood, Buccal Cells, and Fibroblasts From Patients with Inherited Bone Marrow Failure Syndromes.. Blood, 2009, 114, 1083-1083.	0.6	4
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