Inderjeet Dokal

List of Publications by Year
in descending order

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112 papers	11,598 citations	30070 54 h-index	29157 104 g-index
112 all docs	112 docs citations	112 times ranked	8927 citing authors

#	Article	IF	Citations
1	The RNA component of telomerase is mutated in autosomal dominant dyskeratosis congenita. Nature, 2001, 413, 432-435.	27.8	902
2	X-linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolar functions. Nature Genetics, 1998, 19, 32-38.	21.4	867
3	Guidelines for the diagnosis and management of adult aplastic anaemia. British Journal of Haematology, 2016, 172, 187-207.	2.5	539
4	Dyskeratosis congenita in all its forms. British Journal of Haematology, 2000, 110, 768-779.	2.5	487
5	Guidelines for the diagnosis and management of aplastic anaemia. British Journal of Haematology, 2009, 147, 43-70.	2.5	487
6	Disease anticipation is associated with progressive telomere shortening in families with dyskeratosis congenita due to mutations in TERC. Nature Genetics, 2004, 36, 447-449.	21.4	425
7	A human ortholog of archaeal DNA repair protein Hef is defective in Fanconi anemia complementation group M. Nature Genetics, 2005, 37, 958-963.	21.4	395
8	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the ClinicalÂDiagnosis of Inborn Errors of Immunity. Journal of Allergy and Clinical Immunology: in Practice, 2019, 7, 1763-1770.	3.8	381
9	Mutations in dyskeratosis congenita: their impact on telomere length and the diversity of clinical presentation. Blood, 2006, 107, 2680-2685.	1.4	310
10	Mutations in the telomerase component NHP2 cause the premature ageing syndrome dyskeratosis congenita. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 8073-8078.	7.1	294
11	Genetic heterogeneity in autosomal recessive dyskeratosis congenita with one subtype due to mutations in the telomerase-associated protein NOP10. Human Molecular Genetics, 2007, 16, 1619-1629.	2.9	285
12	TINF2 mutations result in very short telomeres: analysis of a large cohort of patients with dyskeratosis congenita and related bone marrow failure syndromes. Blood, 2008, 112, 3594-3600.	1.4	284
13	Association between aplastic anaemia and mutations in telomerase RNA. Lancet, The, 2002, 359, 2168-2170.	13.7	261
14	Dyskeratosis Congenita. Hematology American Society of Hematology Education Program, 2011, 2011, 480-486.	2.5	236
15	Very Short Telomeres in the Peripheral Blood of Patients with X-Linked and Autosomal Dyskeratosis Congenita. Blood Cells, Molecules, and Diseases, 2001, 27, 353-357.	1.4	196
16	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. American Journal of Human Genetics, 2013, 92, 448-453.	6.2	191
17	The Loss of Telomerase Activity in Highly Differentiated CD8+CD28â^'CD27â^' T Cells Is Associated with Decreased Akt (Ser473) Phosphorylation. Journal of Immunology, 2007, 178, 7710-7719.	0.8	185
18	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. Blood Cells, Molecules, and Diseases, 2005, 34, 257-263.	1.4	174

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19	Dyskeratosis Congenita (DC) Registry: identification of new features of DC. British Journal of Haematology, 1998, 103, 990-996.	2.5	165
20	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Journal of Clinical Investigation, 2015, 125, 2151-2160.	8.2	165
21	Disease evolution and outcomes in familial AML with germline CEBPA mutations. Blood, 2015, 126, 1214-1223.	1.4	157
22	Telomerase reverse-transcriptase homozygous mutations in autosomal recessive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. Blood, 2007, 110, 4198-4205.	1.4	155
23	Advances in the understanding of dyskeratosis congenita. British Journal of Haematology, 2009, 145, 164-172.	2.5	141
24	Dyskeratosis congenita, stem cells and telomeres. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 371-379.	3.8	139
25	Engraftment of Hematopoietic Progenitor Cells Transduced with the Fanconi Anemia Group C Gene (FANCC). Human Gene Therapy, 1999, 10, 2337-2346.	2.7	127
26	Fanconi anaemia and leukaemia â€“ clinical and molecular aspects. British Journal of Haematology, 2004, 126, 176-191.	2.5	124
27	Heterozygous telomerase RNA mutations found in dyskeratosis congenita and aplastic anemia reduce telomerase activity via haploinsufficiency. Blood, 2004, 104, 3936-3942.	1.4	122
28	Dyskeratosis Congenita. Seminars in Hematology, 2006, 43, 157-166.	3.4	116
29	Dyskeratosis congenita: telomerase, telomeres and anticipation. Current Opinion in Genetics and Development, 2005, 15, 249-257.	3.3	113
30	Dyskeratosis congenita: Advances in the understanding of the telomerase defect and the role of stem cell transplantation. Pediatric Transplantation, 2007, 11, 584-594.	1.0	109
31	Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia. Human Mutation, 2009, 30, 1567-1573.	2.5	107
32	Inherited bone marrow failure syndromes. Haematologica, 2010, 95, 1236-1240.	3.5	105
33	Revertant Somatic Mosaicism by Mitotic Recombination in Dyskeratosis Congenita. American Journal of Human Genetics, 2012, 90, 426-433.	6.2	97
34	Exome Sequencing Identifies Autosomal-Dominant SRP72 Mutations Associated with Familial Aplasia and Myelodysplasia. American Journal of Human Genetics, 2012, 90, 888-892.	6.2	94
35	Targeted disruption of Dkc1, the gene mutated in X-linked dyskeratosis congenita, causes embryonic lethality in mice. Oncogene, 2002, 21, 7740-7744.	5.9	93
36	Inherited aplastic anaemias/bone marrow failure syndromes. Blood Reviews, 2008, 22, 141-153.	5.7	92

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37	Dyskeratosis congenita: its link to telomerase and aplastic anaemia. Blood Reviews, 2003, 17, 217-225.	5.7	90
38	Mutations in C16orf57 and normal-length telomeres unify a subset of patients with dyskeratosis congenita, poikiloderma with neutropenia and Rothmundâ€“Thomson syndrome. Human Molecular Genetics, 2010, 19, 4453-4461.	2.9	87
39	Bone marrow transplantation for Î²-thalassaemia major: the UK experience in two paediatric centres. British Journal of Haematology, 2003, 120, 289-295.	2.5	85
40	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. American Journal of Human Genetics, 2016, 99, 115-124.	6.2	85
41	Germline NPM1 mutations lead to altered rRNA 2â€²-O-methylation and cause dyskeratosis congenita. Nature Genetics, 2019, 51, 1518-1529.	21.4	84
42	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nature Communications, 2020, 11, 1044.	12.8	81
43	Differences in Disease Severity but Similar Telomere Lengths in Genetic Subgroups of Patients with Telomerase and Shelterin Mutations. PLoS ONE, 2011, 6, e24383.	2.5	79
44	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. Human Genetics, 2001, 108, 299-303.	3.8	77
45	Further delineation of the congenital form of X-linked dyskeratosis congenita (Hoyeraal-Hreidarsson) Tj ETQq1 1 0.784314 rgBT /Over	2.7	70
46	Emberger syndromeâ€”Primary lymphedema with myelodysplasia: Report of seven new cases. American Journal of Medical Genetics, Part A, 2010, 152A, 2287-2296.	1.2	70
47	A novel DKC1 mutation, severe combined immunodeficiency (T+ B- NK- SCID) and bone marrow transplantation in an infant with Hoyeraal-Hreidarsson syndrome. British Journal of Haematology, 2002, 119, 765-768.	2.5	69
48	Dyskeratosis congenita: an inherited bone marrow failure syndrome. British Journal of Haematology, 1996, 92, 775-779.	2.5	67
49	Dyskeratosis congenita: The first NIH clinical research workshop. Pediatric Blood and Cancer, 2009, 53, 520-523.	1.5	66
50	Dyskeratosis Congenita: A historical perspective. Mechanisms of Ageing and Development, 2008, 129, 48-59.	4.6	65
51	Marked genetic heterogeneity in familial myelodysplasia/acute myeloid leukaemia. British Journal of Haematology, 2012, 158, 242-248.	2.5	65
52	Aberrant 3â€² oligoadenylation of spliceosomal U6 small nuclear RNA in poikiloderma with neutropenia. Blood, 2013, 121, 1028-1038.	1.4	65
53	Clinical utility gene card for: Dyskeratosis congenita â€“ update 2015. European Journal of Human Genetics, 2015, 23, 558-558.	2.8	63
54	Adult onset of acute myeloid leukaemia (M6) in patients with Shwachmanâ€Diamond syndrome. British Journal of Haematology, 1997, 99, 171-173.	2.5	59

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55	Functional characterization of novel telomerase RNA (TERC) mutations in patients with diverse clinical and pathological presentations. Haematologica, 2007, 92, 1013-1020.	3.5	58
56	ERCC6L2 Mutations Link a Distinct Bone-Marrow-Failure Syndrome to DNA Repair and Mitochondrial Function. American Journal of Human Genetics, 2014, 94, 246-256.	6.2	58
57	Haematological recovery in dyskeratosis congenita patients treated with danazol. British Journal of Haematology, 2013, 162, 854-856.	2.5	49
58	Triallelic and epigenetic-like inheritance in human disorders of telomerase. Blood, 2015, 126, 176-184.	1.4	49
59	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. Leukemia, 2018, 32, 2502-2507.	7.2	48
60	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica, 2008, 93, 943-944.	3.5	43
61	Novel mutations and polymorphisms in the Fanconi anemia group C gene. Human Mutation, 1996, 8, 140-148.	2.5	42
62	A disease of premature ageing. Lancet, The, 2001, 358, S27.	13.7	42
63	Exome sequencing identifies MPL as a causative gene in familial aplastic anemia. Haematologica, 2012, 97, 524-528.	3.5	42
64	Dyskeratosis congenita: molecular insights into telomerase function, ageing and cancer. Expert Reviews in Molecular Medicine, 2004, 6, 1-23.	3.9	41
65	Dyskeratosis Congenita Is a Chromosomal Instability Disorder. Leukemia and Lymphoma, 1994, 15, 1-7.	1.3	40
66	Dyskeratosis Congenita: A Disorder of Defective Telomere Maintenance?. International Journal of Hematology, 2005, 82, 184-189.	1.6	40
67	Genome instability is a consequence of transcription deficiency in patients with bone marrow failure harboring biallelic ERCC6L2 variants. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 7777-7782.	7.1	37
68	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. Haematologica, 2016, 101, 1180-1189.	3.5	34
69	Chromosomal breakage analysis in dyskeratosis congenita peripheral blood lymphocytes. British Journal of Haematology, 1998, 102, 1162-1164.	2.5	33
70	Two brothers with findings resembling congenital intrauterine infection-like syndrome (pseudo-TORCH syndrome). American Journal of Medical Genetics Part A, 2003, 120A, 261-265.	2.4	32
71	The genetics of Fanconi's anaemia. Best Practice and Research in Clinical Haematology, 2000, 13, 407-425.	1.7	31
72	Fanconi's anaemia and related bone marrow failure syndromes. British Medical Bulletin, 2006, 77-78, 37-53.	6.9	31

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73	Exogenous TERC alone can enhance proliferative potential, telomerase activity and telomere length in lymphocytes from dyskeratosis congenita patients. British Journal of Haematology, 2009, 144, 771-781.	2.5	31
74	Dyskeratosis congenita and the DNA damage response. British Journal of Haematology, 2011, 153, 634-643.	2.5	29
75	A mutation in a functional Sp1 binding site of the telomerase RNA gene (hTERC) promoter in a patient with Paroxysmal Nocturnal Haemoglobinuria. BMC Hematology, 2004, 4, 3.	2.6	28
76	Ataxia and pancytopenia caused by a mutation in TINF2. Human Genetics, 2008, 124, 507-513.	3.8	26
77	Clinical utility gene card for: Dyskeratosis congenita. European Journal of Human Genetics, 2011, 19, 3-4.	2.8	24
78	Severe Variant of Xâ€linked Dyskeratosis Congenita (Hoyeraalâ€Hreidarsson Syndrome) Causes Significant Enterocolitis in Early Infancy. Journal of Pediatric Gastroenterology and Nutrition, 2009, 49, 359-363.	1.8	23
79	Single-Molecule Analysis of the Human Telomerase RNAÂ·Dyskerin Interaction and the Effect of Dyskeratosis Congenita Mutations. Biochemistry, 2009, 48, 10858-10865.	2.5	21
80	Total Deletion of in Vivo Telomere Elongation Capacity: An Ambitious but Possibly Ultimate Cure for All Ageâ€Related Human Cancers. Annals of the New York Academy of Sciences, 2004, 1019, 147-170.	3.8	20
81	Expanding the phenotypic and genetic spectrum of radioulnar synostosis associated hematological disease. Haematologica, 2018, 103, e284-e287.	3.5	17
82	Inherited aplastic anaemia. The Hematology Journal, 2003, 4, 3-9.	1.4	17
83	Limbal stem cell deficiency in patients with inherited stem cell disorder of dyskeratosis congenita. International Ophthalmology, 2012, 32, 615-622.	1.4	16
84	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenitaâ€“like phenotypes. Blood, 2018, 132, 1349-1353.	1.4	16
85	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. Haematologica, 2017, 102, e293-e296.	3.5	15
86	Severe aplastic anemia including FanconiÊ¼s anemia and dyskeratosis congenita. Current Opinion in Hematology, 1996, 3, 453-460.	2.5	13
87	Inherited bone marrow failure syndromes. Hematology, 2015, 20, 433-434.	1.5	13
88	High-throughput STELA provides a rapid test for the diagnosis of telomere biology disorders. Human Genetics, 2021, 140, 945-955.	3.8	12
89	Peripheral neuropathyâ€”a novel finding in dyskeratosis congenita. European Journal of Paediatric Neurology, 2005, 9, 85-89.	1.6	8
90	Circulating haematopoietic progenitors are differentially reduced amongst subtypes of dyskeratosis congenita. British Journal of Haematology, 2008, 140, 719-722.	2.5	8

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91	Dyskeratosis congenita: a disorder of telomerase deficiency and its relationship to other diseases. Expert Review of Dermatology, 2006, 1, 463-479.	0.3	7
92	Hepatitis-associated Aplastic Anemia Presenting as a Familial Bone Marrow Failure Syndrome. Journal of Pediatric Hematology/Oncology, 2009, 31, 884-887.	0.6	7
93	Fanconi anemia is a highly penetrant cancer susceptibility syndrome. Haematologica, 2008, 93, 486-8.	3.5	7
94	Prenatal and postnatal growth retardation, microcephaly, developmental delay, and pigmentation abnormalities: Naegeli syndrome, dyskeratosis congenita, poikiloderma Clericuzio type, or separate entity?. European Journal of Medical Genetics, 2011, 54, 231-235.	1.3	6
95	Understanding aplastic anaemia/bone-marrow failure syndromes. Paediatrics and Child Health (United) Tj ETQq1 1 0.784314 rgBT /Ov	0.4	5
96	Inherited bone marrow failure syndromes. Journal of Hematopathology, 2011, 4, 53-60.	0.4	5
97	Heightened DNA damage response impairs hematopoiesis in Fanconi anemia. Haematologica, 2012, 97, 1117-1117.	3.5	5
98	TGF-Î² Pathway Inhibition Signals New Hope for Fanconi Anemia. Cell Stem Cell, 2016, 18, 567-568.	11.1	5
99	The Inherited Bone Marrow Failure Syndromes: Fanconi Anemia, Dyskeratosis Congenita and Diamondâ€Blackfan Anemia. Reviews in Clinical and Experimental Hematology, 2000, 4, 183-215.	0.1	4
100	Detection of hereditary haemochromatosis in an HLA-identical pedigree showing discordance between HLA class I genes and the disease locus. Human Genetics, 1991, 88, 209-14.	3.8	3
101	Telomerase dysfunction and dyskeratosis congenita. Cytotechnology, 2004, 45, 13-22.	1.6	3
102	A frameshift variant in specificity protein 1 triggers superactivation of Sp1-mediated transcription in familial bone marrow failure. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 17151-17155.	7.1	2
103	Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. Blood, 2021, 138, 864-864.	1.4	2
104	Dyskeratosis Congenita. , 2014, , 267-280.		1
105	British Committee for Standards in Haematology guidelines for aplastic anemia: Single centre retrospective review finds no compelling evidence for the recommended higher platelet count threshold of 20ÂÃ—Â10⁹/L â€•<scp>RESPONSE</scp> to Yan etÂal. British Journal of Haematology, 2018, 182, 286-287.	2.5	1
106	Genome-wide whole-blood transcriptome profiling across inherited bone marrow failure subtypes. Blood Advances, 2021, 5, 5360-5371.	5.2	1
107	Detection of implanted splenic tissue using 99m-Technetium-labelled heat-damaged autologous red cells. British Journal of Haematology, 2002, 118, 2-2.	2.5	0
108	Diagnostic radioisotopes in haematology. , 2006, , 357-378.		0

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109	Dissecting â€œstressâ€•in Fanconi anemia. Blood, 2008, 111, 1756-1757.	1.4	0
110	Low Dose Alemtuzumab Achieves Long-Term Engraftment with Low Level Mixed Chimerism in Related Haemopoietic Stem Cell Transplantation for Haemoglobinopathies. Blood, 2008, 112, 3304-3304.	1.4	0
111	Full Donor Engraftment without Chronic GvHD Following HLA-Matched Sibling Donor Haemopoietic Stem Cell Transplantation for Diamond Blackfan Anaemia. Blood, 2008, 112, 4394-4394.	1.4	0
112	Overlap of Syndromes Associated with Myelodysplasia and Leukemia. Blood, 2014, 124, SCI-32-SCI-32.	1.4	0