

# Inderjeet Dokal

## List of Publications by Year in descending order

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112  
papers

11,598  
citations

29994

54  
h-index

30010

103  
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. <i>Nature</i> , 2001, 413, 432-435.	13.7	902
2	X-linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolar functions. <i>Nature Genetics</i> , 1998, 19, 32-38.	9.4	867
3	Guidelines for the diagnosis and management of adult aplastic anaemia. <i>British Journal of Haematology</i> , 2016, 172, 187-207.	1.2	539
4	Dyskeratosis congenita in all its forms. <i>British Journal of Haematology</i> , 2000, 110, 768-779.	1.2	487
5	Guidelines for the diagnosis and management of aplastic anaemia. <i>British Journal of Haematology</i> , 2009, 147, 43-70.	1.2	487
6	Disease anticipation is associated with progressive telomere shortening in families with dyskeratosis congenita due to mutations in TERC. <i>Nature Genetics</i> , 2004, 36, 447-449.	9.4	425
7	A human ortholog of archaeal DNA repair protein Hef is defective in Fanconi anemia complementation group M. <i>Nature Genetics</i> , 2005, 37, 958-963.	9.4	395
8	The European Society for Immunodeficiencies (ESID) Registry Working Definitions for the Clinical Diagnosis of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2019, 7, 1763-1770.	2.0	381
9	Mutations in dyskeratosis congenita: their impact on telomere length and the diversity of clinical presentation. <i>Blood</i> , 2006, 107, 2680-2685.	0.6	310
10	Mutations in the telomerase component NHP2 cause the premature ageing syndrome dyskeratosis congenita. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 8073-8078.	3.3	294
11	Genetic heterogeneity in autosomal recessive dyskeratosis congenita with one subtype due to mutations in the telomerase-associated protein NOP10. <i>Human Molecular Genetics</i> , 2007, 16, 1619-1629.	1.4	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. <i>Blood</i> , 2008, 112, 3594-3600.	0.6	284
13	Association between aplastic anaemia and mutations in telomerase RNA. <i>Lancet, The</i> , 2002, 359, 2168-2170.	6.3	261
14	Dyskeratosis Congenita. <i>Hematology American Society of Hematology Education Program</i> , 2011, 2011, 480-486.	0.9	236
15	Very Short Telomeres in the Peripheral Blood of Patients with X-Linked and Autosomal Dyskeratosis Congenita. <i>Blood Cells, Molecules, and Diseases</i> , 2001, 27, 353-357.	0.6	196
16	Constitutional Mutations in RTEL1 Cause Severe Dyskeratosis Congenita. <i>American Journal of Human Genetics</i> , 2013, 92, 448-453.	2.6	191
17	The Loss of Telomerase Activity in Highly Differentiated CD8+CD28 <sup>hi</sup> CD27 <sup>hi</sup> T Cells Is Associated with Decreased Akt (Ser473) Phosphorylation. <i>Journal of Immunology</i> , 2007, 178, 7710-7719.	0.4	185
18	Mutations in the reverse transcriptase component of telomerase (TERT) in patients with bone marrow failure. <i>Blood Cells, Molecules, and Diseases</i> , 2005, 34, 257-263.	0.6	174

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19	Dyskeratosis Congenita (DC) Registry: identification of new features of DC. British Journal of Haematology, 1998, 103, 990-996.	1.2	165
20	Poly(A)-specific ribonuclease deficiency impacts telomere biology and causes dyskeratosis congenita. Journal of Clinical Investigation, 2015, 125, 2151-2160.	3.9	165
21	Disease evolution and outcomes in familial AML with germline CEBPA mutations. Blood, 2015, 126, 1214-1223.	0.6	157
22	Telomerase reverse-transcriptase homozygous mutations in autosomal recessive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. Blood, 2007, 110, 4198-4205.	0.6	155
23	Advances in the understanding of dyskeratosis congenita. British Journal of Haematology, 2009, 145, 164-172.	1.2	141
24	Dyskeratosis congenita, stem cells and telomeres. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 371-379.	1.8	139
25	Engraftment of Hematopoietic Progenitor Cells Transduced with the Fanconi Anemia Group C Gene (FANCC). Human Gene Therapy, 1999, 10, 2337-2346.	1.4	127
26	Fanconi anaemia and leukaemia - clinical and molecular aspects. British Journal of Haematology, 2004, 126, 176-191.	1.2	124
27	Heterozygous telomerase RNA mutations found in dyskeratosis congenita and aplastic anemia reduce telomerase activity via haploinsufficiency. Blood, 2004, 104, 3936-3942.	0.6	122
28	Dyskeratosis Congenita. Seminars in Hematology, 2006, 43, 157-166.	1.8	116
29	Dyskeratosis congenita: telomerase, telomeres and anticipation. Current Opinion in Genetics and Development, 2005, 15, 249-257.	1.5	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.	0.5	109
31	Defining the pathogenic role of telomerase mutations in myelodysplastic syndrome and acute myeloid leukemia. Human Mutation, 2009, 30, 1567-1573.	1.1	107
32	Inherited bone marrow failure syndromes. Haematologica, 2010, 95, 1236-1240.	1.7	105
33	Revertant Somatic Mosaicism by Mitotic Recombination in Dyskeratosis Congenita. American Journal of Human Genetics, 2012, 90, 426-433.	2.6	97
34	Exome Sequencing Identifies Autosomal-Dominant SRP72 Mutations Associated with Familial Aplasia and Myelodysplasia. American Journal of Human Genetics, 2012, 90, 888-892.	2.6	94
35	Targeted disruption of Dkc1, the gene mutated in X-linked dyskeratosis congenita, causes embryonic lethality in mice. Oncogene, 2002, 21, 7740-7744.	2.6	93
36	Inherited aplastic anaemias/bone marrow failure syndromes. Blood Reviews, 2008, 22, 141-153.	2.8	92

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37	Dyskeratosis congenita: its link to telomerase and aplastic anaemia. <i>Blood Reviews</i> , 2003, 17, 217-225.	2.8	90
38	Mutations in C16orf57 and normal-length telomeres unify a subset of patients with dyskeratosis congenita, poikiloderma with neutropenia and Rothmund-Thomson syndrome. <i>Human Molecular Genetics</i> , 2010, 19, 4453-4461.	1.4	87
39	Bone marrow transplantation for $\beta^2$ -thalassaemia major: the UK experience in two paediatric centres. <i>British Journal of Haematology</i> , 2003, 120, 289-295.	1.2	85
40	DNAJC21 Mutations Link a Cancer-Prone Bone Marrow Failure Syndrome to Corruption in 60S Ribosome Subunit Maturation. <i>American Journal of Human Genetics</i> , 2016, 99, 115-124.	2.6	85
41	Germline NPM1 mutations lead to altered rRNA 2'-O-methylation and cause dyskeratosis congenita. <i>Nature Genetics</i> , 2019, 51, 1518-1529.	9.4	84
42	The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. <i>Nature Communications</i> , 2020, 11, 1044.	5.8	81
43	Differences in Disease Severity but Similar Telomere Lengths in Genetic Subgroups of Patients with Telomerase and Shelterin Mutations. <i>PLoS ONE</i> , 2011, 6, e24383.	1.1	79
44	Identification of novel DKC1 mutations in patients with dyskeratosis congenita: implications for pathophysiology and diagnosis. <i>Human Genetics</i> , 2001, 108, 299-303.	1.8	77
45	Further delineation of the congenital form of X-linked dyskeratosis congenita (Hoyeraal-Hreidarsson) Tj ETQq1 1 0.784314 rgBT /Over	1.3	70
46	Emberger syndrome—Primary lymphedema with myelodysplasia: Report of seven new cases. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2287-2296.	0.7	70
47	A novel DKC1 mutation, severe combined immunodeficiency (T+ B- NK- SCID) and bone marrow transplantation in an infant with Hoyeraal-Hreidarsson syndrome. <i>British Journal of Haematology</i> , 2002, 119, 765-768.	1.2	69
48	Dyskeratosis congenita: an inherited bone marrow failure syndrome. <i>British Journal of Haematology</i> , 1996, 92, 775-779.	1.2	67
49	Dyskeratosis congenita: The first NIH clinical research workshop. <i>Pediatric Blood and Cancer</i> , 2009, 53, 520-523.	0.8	66
50	Dyskeratosis Congenita: A historical perspective. <i>Mechanisms of Ageing and Development</i> , 2008, 129, 48-59.	2.2	65
51	Marked genetic heterogeneity in familial myelodysplasia/acute myeloid leukaemia. <i>British Journal of Haematology</i> , 2012, 158, 242-248.	1.2	65
52	Aberrant 3' oligoadenylation of spliceosomal U6 small nuclear RNA in poikiloderma with neutropenia. <i>Blood</i> , 2013, 121, 1028-1038.	0.6	65
53	Clinical utility gene card for: Dyskeratosis congenita — update 2015. <i>European Journal of Human Genetics</i> , 2015, 23, 558-558.	1.4	63
54	Adult onset of acute myeloid leukaemia (M6) in patients with Shwachman-Diamond syndrome. <i>British Journal of Haematology</i> , 1997, 99, 171-173.	1.2	59

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55	Functional characterization of novel telomerase RNA (TERC) mutations in patients with diverse clinical and pathological presentations. <i>Haematologica</i> , 2007, 92, 1013-1020.	1.7	58
56	ERCC6L2 Mutations Link a Distinct Bone-Marrow-Failure Syndrome to DNA Repair and Mitochondrial Function. <i>American Journal of Human Genetics</i> , 2014, 94, 246-256.	2.6	58
57	Haematological recovery in dyskeratosis congenita patients treated with danazol. <i>British Journal of Haematology</i> , 2013, 162, 854-856.	1.2	49
58	Triallelic and epigenetic-like inheritance in human disorders of telomerase. <i>Blood</i> , 2015, 126, 176-184.	0.6	49
59	GATA2 monoallelic expression underlies reduced penetrance in inherited GATA2-mutated MDS/AML. <i>Leukemia</i> , 2018, 32, 2502-2507.	3.3	48
60	Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. <i>Haematologica</i> , 2008, 93, 943-944.	1.7	43
61	Novel mutations and polymorphisms in the Fanconi anemia group C gene. , 1996, 8, 140-148.		42
62	A disease of premature ageing. <i>Lancet, The</i> , 2001, 358, S27.	6.3	42
63	Exome sequencing identifies MPL as a causative gene in familial aplastic anemia. <i>Haematologica</i> , 2012, 97, 524-528.	1.7	42
64	Dyskeratosis congenita: molecular insights into telomerase function, ageing and cancer. <i>Expert Reviews in Molecular Medicine</i> , 2004, 6, 1-23.	1.6	41
65	Dyskeratosis Congenita Is a Chromosomal Instability Disorder. <i>Leukemia and Lymphoma</i> , 1994, 15, 1-7.	0.6	40
66	Dyskeratosis Congenita: A Disorder of Defective Telomere Maintenance?. <i>International Journal of Hematology</i> , 2005, 82, 184-189.	0.7	40
67	Genome instability is a consequence of transcription deficiency in patients with bone marrow failure harboring biallelic <i>ERCC6L2</i> variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 7777-7782.	3.3	37
68	Marked overlap of four genetic syndromes with dyskeratosis congenita confounds clinical diagnosis. <i>Haematologica</i> , 2016, 101, 1180-1189.	1.7	34
69	Chromosomal breakage analysis in dyskeratosis congenita peripheral blood lymphocytes. <i>British Journal of Haematology</i> , 1998, 102, 1162-1164.	1.2	33
70	Two brothers with findings resembling congenital intrauterine infection-like syndrome (pseudo-TORCH syndrome). <i>American Journal of Medical Genetics Part A</i> , 2003, 120A, 261-265.	2.4	32
71	The genetics of Fanconi's anaemia. <i>Best Practice and Research in Clinical Haematology</i> , 2000, 13, 407-425.	0.7	31
72	Fanconi's anaemia and related bone marrow failure syndromes. <i>British Medical Bulletin</i> , 2006, 77-78, 37-53.	2.7	31

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73	Exogenous <i>TERC</i> alone can enhance proliferative potential, telomerase activity and telomere length in lymphocytes from dyskeratosis congenita patients. <i>British Journal of Haematology</i> , 2009, 144, 771-781.	1.2	31
74	Dyskeratosis congenita and the DNA damage response. <i>British Journal of Haematology</i> , 2011, 153, 634-643.	1.2	29
75	A mutation in a functional Sp1 binding site of the telomerase RNA gene (hTERC) promoter in a patient with Paroxysmal Nocturnal Haemoglobinuria. <i>BMC Hematology</i> , 2004, 4, 3.	2.6	28
76	Ataxia and pancytopenia caused by a mutation in TINF2. <i>Human Genetics</i> , 2008, 124, 507-513.	1.8	26
77	Clinical utility gene card for: Dyskeratosis congenita. <i>European Journal of Human Genetics</i> , 2011, 19, 3-4.	1.4	24
78	Severe Variant of X-linked Dyskeratosis Congenita (Hoyeraal-Hreidarsson Syndrome) Causes Significant Enterocolitis in Early Infancy. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2009, 49, 359-363.	0.9	23
79	Single-Molecule Analysis of the Human Telomerase RNA-Dyskerin Interaction and the Effect of Dyskeratosis Congenita Mutations. <i>Biochemistry</i> , 2009, 48, 10858-10865.	1.2	21
80	Total Deletion of <i>in Vivo</i> Telomere Elongation Capacity: An Ambitious but Possibly Ultimate Cure for All Age-Related Human Cancers. <i>Annals of the New York Academy of Sciences</i> , 2004, 1019, 147-170.	1.8	20
81	Expanding the phenotypic and genetic spectrum of radioulnar synostosis associated hematological disease. <i>Haematologica</i> , 2018, 103, e284-e287.	1.7	17
82	Inherited aplastic anaemia. <i>The Hematology Journal</i> , 2003, 4, 3-9.	2.0	17
83	Limbic stem cell deficiency in patients with inherited stem cell disorder of dyskeratosis congenita. <i>International Ophthalmology</i> , 2012, 32, 615-622.	0.6	16
84	Homozygous OB-fold variants in telomere protein TPP1 are associated with dyskeratosis congenita-like phenotypes. <i>Blood</i> , 2018, 132, 1349-1353.	0.6	16
85	Myelodysplasia and liver disease extend the spectrum of RTEL1 related telomeropathies. <i>Haematologica</i> , 2017, 102, e293-e296.	1.7	15
86	Severe aplastic anemia including Fanconi's anemia and dyskeratosis congenita. <i>Current Opinion in Hematology</i> , 1996, 3, 453-460.	1.2	13
87	Inherited bone marrow failure syndromes. <i>Hematology</i> , 2015, 20, 433-434.	0.7	13
88	High-throughput STELA provides a rapid test for the diagnosis of telomere biology disorders. <i>Human Genetics</i> , 2021, 140, 945-955.	1.8	12
89	Peripheral neuropathy—a novel finding in dyskeratosis congenita. <i>European Journal of Paediatric Neurology</i> , 2005, 9, 85-89.	0.7	8
90	Circulating haematopoietic progenitors are differentially reduced amongst subtypes of dyskeratosis congenita. <i>British Journal of Haematology</i> , 2008, 140, 719-722.	1.2	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.	0.3	7
93	Fanconi anemia is a highly penetrant cancer susceptibility syndrome. Haematologica, 2008, 93, 486-8.	1.7	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.	0.7	6
95	Understanding aplastic anaemia/bone-marrow failure syndromes. Paediatrics and Child Health (United Kingdom), 2011, 47, 102-105.	0.2	5
96	Inherited bone marrow failure syndromes. Journal of Hematopathology, 2011, 4, 53-60.	0.2	5
97	Heightened DNA damage response impairs hematopoiesis in Fanconi anemia. Haematologica, 2012, 97, 1117-1117.	1.7	5
98	TGF- $\beta$ 2 Pathway Inhibition Signals New Hope for Fanconi Anemia. Cell Stem Cell, 2016, 18, 567-568.	5.2	5
99	The Inherited Bone Marrow Failure Syndromes: Fanconi Anemia, Dyskeratosis Congenita and Diamond-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.	1.8	3
101	Telomerase dysfunction and dyskeratosis congenita. Cytotechnology, 2004, 45, 13-22.	0.7	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.	3.3	2
103	Multinational Study on the Clinical and Genetic Features of the ERCC6L2-Disease. Blood, 2021, 138, 864-864.	0.6	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 $20 \times 10^9/L$ to $30 \times 10^9/L$ . British Journal of Haematology, 2018, 182, 286-287.	1.2	1
106	Genome-wide whole-blood transcriptome profiling across inherited bone marrow failure subtypes. Blood Advances, 2021, 5, 5360-5371.	2.5	1
107	Detection of implanted splenic tissue using $^{99m}\text{Tc}$ -labelled heat-damaged autologous red cells. British Journal of Haematology, 2002, 118, 2-2.	1.2	0
108	Diagnostic radioisotopes in haematology. , 2006, , 357-378.		0

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109	Dissecting "stress" in Fanconi anemia. Blood, 2008, 111, 1756-1757.	0.6	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.	0.6	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.	0.6	0
112	Overlap of Syndromes Associated with Myelodysplasia and Leukemia. Blood, 2014, 124, SCI-32-SCI-32.	0.6	0